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Patentanmeldung Nr. Patent application No. Demande de brevet n° ..

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Der Präsident des Europäischen Patentamts;
Im Auftrag

For the President of the European Patent Office
Le Président de l'Office européen des brevets
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R C van Dijk

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Single nucleotide polymorphisms sensitively predicting adverse drug reactions (ADR) and drug efficacy

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Single Nucleotide Polymorphisms sensitively predicting Adverse Drug Reactions (ADR) and Drug Efficacy

Technical Field

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This invention relates to genetic polymorphisms useful for assessing cardiovascular risks in humans, including, but not limited to, atherosclerosis, ischemia/reperfusion, hypertension, restenosis, arterial inflammation, myocardial infarction, and stroke. In addition it relates to genetic polymorphisms useful for assessing the response to lipid lowering drug therapy. Specifically, the present invention identifies and describes gene variations which are individually present in humans with cardiovascular disease states, relate to humans with normal, or non-cardiovascular disease states, and/or in response to medications relevant to cardiovascular disease. Further, the present invention provides methods for the identification and therapeutic use of compounds as treatments of cardiovascular disease. Moreover, the present invention provides methods for the diagnostic monitoring of patients undergoing clinical evaluation for the treatment of cardiovascular disease, and for monitoring the efficacy of compounds in clinical trials. Still further, the present invention provides methods to use gene variations to predict personal medication schemes omitting adverse drug reactions and allowing an adjustment of the drug dose to achieve maximum benefit for the patient. Additionally, the present invention describes methods for the diagnostic evaluation and prognosis of various cardiovascular diseases, and for the identification of subjects exhibiting a predisposition to such conditions.

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Background of the Invention

Cardiovascular disease is a major health risk throughout the industrialized world.

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Cardiovascular diseases include but are not limited by the following disorders of the heart and the vascular system: congestive heart failure, myocardial infarction, atherosclerosis, ischemic diseases of the heart, coronary heart disease, all kinds of

atrial and ventricular arrhythmias, hypertensive vascular diseases and peripheral vascular diseases.

- 5 Heart failure is defined as a pathophysiologic state in which an abnormality of cardiac function is responsible for the failure of the heart to pump blood at a rate commensurate with the requirement of the metabolizing tissue. It includes all forms of pumping failure such as high-output and low-output, acute and chronic, right-sided or left-sided, systolic or diastolic, independent of the underlying cause.
- 10 Myocardial infarction (MI) is generally caused by an abrupt decrease in coronary blood flow that follows a thrombotic occlusion of a coronary artery previously narrowed by arteriosclerosis. MI prophylaxis (primary and secondary prevention) is included as well as the acute treatment of MI and the prevention of complications.
- 15 Ischemic diseases are conditions in which the coronary flow is restricted resulting in an perfusion which is inadequate to meet the myocardial requirement for oxygen. This group of diseases include stable angina, unstable angina and asymptomatic ischemia.
- 20 Arrhythmias include all forms of atrial and ventricular tachyarrhythmias (atrial tachycardia, atrial flutter, atrial fibrillation, atrio-ventricular reentrant tachycardia, preexcitation syndrome, ventricular tachycardia, ventricular flutter, ventricular fibrillation) as well as bradycardic forms of arrhythmias.
- 25 Hypertensive vascular diseases include primary as well as all kinds of secondary arterial hypertension (renal, endocrine, neurogenic, others).
- 30 Peripheral vascular diseases are defined as vascular diseases in which arterial and/or venous flow is reduced resulting in an imbalance between blood supply and tissue oxygen demand. It includes chronic peripheral arterial occlusive disease (PAOD),

acute arterial thrombosis and embolism, inflammatory vascular disorders, Raynaud's phenomenon and venous disorders.

Atherosclerosis, the most prevalent of vascular diseases, is the principal cause of heart attack, stroke, and gangrene of the extremities, and thereby the principal cause of death. Atherosclerosis is a complex disease involving many cell types and molecular factors (for a detailed review, see Ross, 1993, Nature 362: 801-809 and Lusis, A. J., Nature 407, 233-241 (2000)). The process, in normal circumstances a protective response to insults to the endothelium and smooth muscle cells (SMCs) of the wall of the artery, consists of the formation of fibrofatty and fibrous lesions or plaques, preceded and accompanied by inflammation. The advanced lesions of atherosclerosis may occlude the artery concerned, and result from an excessive inflammatory-fibroproliferative response to numerous different forms of insult. For example, shear stresses are thought to be responsible for the frequent occurrence of atherosclerotic plaques in regions of the circulatory system where turbulent blood flow occurs, such as branch points and irregular structures.

The first observable event in the formation of an atherosclerotic plaque occurs when blood-borne monocytes adhere to the vascular endothelial layer and transmigrate through to the sub-endothelial space. Adjacent endothelial cells at the same time produce oxidized low density lipoprotein (LDL). These oxidized LDLs are then taken up in large amounts by the monocytes through scavenger receptors expressed on their surfaces. In contrast to the regulated pathway by which native LDL (nLDL) is taken up by nLDL specific receptors, the scavenger pathway of uptake is not regulated by the monocytes.

These lipid-filled monocytes are called foam cells, and are the major constituent of the fatty streak. Interactions between foam cells and the endothelial and SMCs which surround them lead to a state of chronic local inflammation which can eventually lead to smooth muscle cell proliferation and migration, and the formation of a fibrous

plaque. Such plaques occlude the blood vessel concerned and thus restrict the flow of blood, resulting in ischemia.

Ischemia is a condition characterized by a lack of oxygen supply in tissues of organs due to inadequate perfusion. Such inadequate perfusion can have number of natural causes, including atherosclerotic or restenotic lesions, anemia, or stroke, to name a few. Many medical interventions, such as the interruption of the flow of blood during bypass surgery, for example, also lead to ischemia. In addition to sometimes being caused by diseased cardiovascular tissue, ischemia may sometimes affect cardiovascular tissue, such as in ischemic heart disease. Ischemia may occur in any organ, however, that is suffering a lack of oxygen supply.

The most common cause of ischemia in the heart is atherosclerotic disease of epicardial coronary arteries. By reducing the lumen of these vessels, atherosclerosis causes an absolute decrease in myocardial perfusion in the basal state or limits appropriate increases in perfusion when the demand for flow is augmented. Coronary blood flow can also be limited by arterial thrombi, spasm, and, rarely, coronary emboli, as well as by ostial narrowing due to luetic aortitis. Congenital abnormalities, such as anomalous origin of the left anterior descending coronary artery from the pulmonary artery, may cause myocardial ischemia and infarction in infancy, but this cause is very rare in adults. Myocardial ischemia can also occur if myocardial oxygen demands are abnormally increased, as in severe ventricular hypertrophy due to hypertension or aortic stenosis. The latter can be present with angina that is indistinguishable from that caused by coronary atherosclerosis. A reduction in the oxygen-carrying capacity of the blood, as in extremely severe anemia or in the presence of carboxy-hemoglobin, is a rare cause of myocardial ischemia. Not infrequently, two or more causes of ischemia will coexist, such as an increase in oxygen demand due to left ventricular hypertrophy and a reduction in oxygen supply secondary to coronary atherosclerosis.

The foregoing studies are aimed at defining the role of particular gene variations presumed to be involved in the misleading of normal cellular function leading to cardiovascular disease. However, such approaches cannot identify the full panoply of gene variations that are involved in the disease process.

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At present, the only available treatments for cardiovascular disorders are pharmaceutical based medications that are not targeted to an individual's actual defect; examples include angiotensin converting enzyme (ACE) inhibitors and diuretics for hypertension, insulin supplementation for non-insulin dependent

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diabetes mellitus (NIDDM), cholesterol reduction strategies for dyslipidaemia, anti-coagulants, β blockers for cardiovascular disorders and weight reduction strategies for obesity. If targeted treatment strategies were available it might be possible to predict the response to a particular regime of therapy and could markedly increase the effectiveness of such treatment. Although targeted therapy requires accurate

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diagnostic tests for disease susceptibility, once these tests are developed the opportunity to utilize targeted therapy will become widespread. Such diagnostic tests could initially serve to identify individuals at most risk of hypertension and could allow them to make changes in lifestyle or diet that would serve as preventative measures. The benefits associated by coupling the diagnostic tests with a system of

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targeted therapy could include the reduction in dosage of administered drugs and thus the amount of unpleasant side effects suffered by an individual. In more severe cases a diagnostic test may suggest that earlier surgical intervention would be useful in preventing a further deterioration in condition.

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It is an object of the invention to provide genetic diagnosis of predisposition or susceptibility for cardiovascular diseases. Another related object is to provide treatment to reduce or prevent or delay the onset of disease in those predisposed or susceptible to this disease. A further object is to provide means for carrying out this diagnosis.

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Accordingly, a first aspect of the invention provides a method of diagnosis of disease in an individual, said method comprising determining one, various or all genotypes in said individual of the genes listed in the Examples.

- 5 In another aspect, the invention provides a method of identifying an individual predisposed or susceptible to a disease, said method comprising determining one, various or all genotypes in said individual of the genes listed in the Examples.

..... The invention is of advantage in that it enables diagnosis of a disease or of certain
10 disease states via genetic analysis which can yield useable results before onset of disease symptoms, or before onset of severe symptoms. The invention is further of advantage in that it enables diagnosis of predisposition or susceptibility to a disease or of certain disease states via genetic analysis.

- 15 The invention may also be of use in confirming or corroborating the results of other diagnostic methods. The diagnosis of the invention may thus suitably be used either as an isolated technique or in combination with other methods and apparatus for diagnosis, in which latter case the invention provides a further test on which a diagnosis may be assessed.

20 The present invention stems from using allelic association as a method for genotyping individuals; allowing the investigation of the molecular genetic basis for cardiovascular diseases. In a specific embodiment the invention tests for the polymorphisms in the sequences of the listed genes in the Examples. The invention demonstrates a link between this polymorphisms and predispositions to cardiovascular diseases by showing that allele frequencies significantly differ when individuals with "bad" serum lipids are compared to individuals with "good" serum levels. The meaning of "good and bad" serum lipid levels is defined in Table 1a.

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- 30 Certain disease states would benefit, that is to say the suffering of the patient may be reduced or prevented or delayed, by administration of treatment or therapy in

advance of disease appearance; this can be more reliably carried out if advance diagnosis of predisposition or susceptibility to disease can be diagnosed.

Pharmacogenomics and adverse drug reactions

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Adverse drug reactions (ADRs) remain a major clinical problem. A recent meta-analysis suggested that in the USA in 1994, ADRs were responsible for 100 000 deaths, making them between the fourth and sixth commonest cause of death (Lazarou 1998, J. Am. Med. Assoc. 279:1200). Although these figures have been heavily criticized, they emphasize the importance of ADRs. Indeed, there is good evidence that ADRs account for 5% of all hospital admissions and increase the length of stay in hospital by two days at an increased cost of ~\$2500 per patient. ADRs are also one of the commonest causes of drug withdrawal, which has enormous financial implications for the pharmaceutical industry. ADRs, perhaps fortunately, only affect a minority of those taking a particular drug. Although factors that determine susceptibility are unclear in most cases, there is increasing interest in the role of genetic factors. Indeed, the role of inheritable variations in predisposing patients to ADRs has been appreciated since the late 1950s and early 1960s through the discovery of deficiencies in enzymes such as pseudocholinesterase (butyrylcholinesterase) and glucose-6-phosphate dehydrogenase (G6PD). More recently, with the first draft of the human genome just completed, there has been renewed interest in this area with the introduction of terms such as pharmacogenomics and toxicogenomics. Essentially, the aim of pharmacogenomics is to produce personalized medicines, whereby administration of the drug class and dosage is tailored to an individual genotype. Thus, the term pharmacogenomics embraces both efficacy and toxicity.

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The 3-hydroxy-3-methylglutaryl coenzyme A (HMG-CoA) reductase inhibitors ("statins") specifically inhibit the enzyme HMG-CoA reductase which catalyzes the rate limiting step in cholesterol biosynthesis. These drugs are effective in reducing the primary and secondary risk of coronary artery disease and coronary events, such

as heart attack, in middle-aged and older men and women, in both diabetic and non-diabetic patients, and are often prescribed for patients with hyperlipidemia. Statins used in secondary prevention of coronary artery or heart disease significantly reduce the risk of stroke, total mortality and morbidity and attacks of myocardial ischemia; 5 the use of statins is also associated with improvements in endothelial and fibrinolytic functions and decreased platelet thrombus formation.

The tolerability of these drugs during long term administration is an important issue. Adverse reactions involving skeletal muscle are not uncommon, and sometimes 10 serious adverse reactions involving skeletal muscle such as myopathy and rhabdomyolysis may occur, requiring discontinuation of the drug. In addition an increase in serum creatine kinase (CK) may be a sign of a statin related adverse event. The extend of such adverse events can be read from the extend of the CK level increase (as compared to the upper limit of normal [ULN]).

15 Occasionally arthralgia, alone or in association with myalgia, has been reported. Also an elevation of liver transaminases has been associated with statin administration.

It was shown that the drug response to statin therapy is a class effects, i.e. all known 20 and presumably also all so far undiscovered statins share the same beneficial and harmful effects (Ucar, M. et al., Drug Safety 2000, 22:441). It follows that the discovery of diagnostic tools to predict the drug response to a single statin will also be of aid to guide therapy with other statins.

25 The present invention provides diagnostic tests to predict the patient's individual response to statin therapy. Such responses include, but are not limited by the extent of adverse drug reactions, the level of lipid lowering or the drug's influence on disease states. Those diagnostic tests may predict the response to statin therapy either alone or in combination with another diagnostic test or another drug regimen.

Detailed Description of the Invention

The present invention is based at least in part on the discovery that a specific allele of a polymorphic region of a so called "candidate gene" (as defined below) is associated
5 with CVD or drug response.

For the present invention the following candidate genes were analyzed:

-Genes found to be expressed in cardiac tissue (Hwang et al., Circulation 1997,
96:4146-4203).

10 -Genes from the following metabolic pathways and their regulatory elements:

Lipid metabolism

Numerous studies have shown a connection between serum lipid levels and
15 cardiovascular diseases. Candidate genes falling into this group include but are not limited by genes of the cholesterol pathway, apolipoproteins and their modifying factors.

Coagulation

20 Ischemic diseases of the heart and in particular myocardial infarction may be caused by a thrombotic occlusion. Genes falling into this group include all genes of the coagulation cascade and their regulatory elements.

25 Inflammation

Complications of atherosclerosis are the most common causes of death in Western societies. In broad outline atherosclerosis can be considered to be a form of chronic inflammation resulting from interaction modified lipoproteins, monocyte-derived
30 macrophages, T cells, and the normal cellular elements of the arterial wall. This inflammatory process can ultimately lead to the development of complex lesions, or

plaques, that protrude into the arterial lumen. Finally plaque rupture and thrombosis result in the acute clinical complications of myocardial infarction and stroke (Glass et al., Cell 2001, 104:503-516).

- 5 It follows that all genes related to inflammatory processes, including but not limited by cytokines, cytokine receptors and cell adhesion molecules are candidate genes for CVD.

Glucose and energy metabolism

- 10 As glucose and energy metabolism is interdependent with the metabolism of lipids (see above) also the former pathways contain candidate genes. Energy metabolism in general also relates to obesity, which is an independent risk factor for CVD (Melanson et al., Cardiol Rev 2001 9:202-207). In addition high blood glucose levels
15 are associated with many microvascular and macrovascular complications and may therefore affect an individuals disposition to CVD (Duckworth, Curr Atheroscler Rep 2001, 3:383-391).

Hypertension

- 20 As hypertension is an independent risk factor for CVD, also genes that are involved in the regulation of systolic and diastolic blood pressure affect an individuals risk for CVD (Safar, Curr Opin Cardiol 2000, 15:258-263). Interestingly hypertension and diabetes (see above) appear to be interdependent, since hypertension is approximately twice as frequent in patients with diabetes compared with patients without the disease. Conversely, recent data suggest that hypertensive persons are more predisposed to the development of diabetes than are normotensive persons
25 (Sowers et al., Hypertension 2001, 37:1053-1059).

Genes related to drug response

Those genes include metabolic pathways involved in the absorption, distribution, metabolism, excretion and toxicity (ADMET) of drugs. Prominent members of this group are the cytochrome P450 proteins which catalyze many reactions involved in drug metabolism.

Unclassified genes

As stated above, the mechanisms that lead to cardiovascular diseases or define the patient's individual response to drugs are not completely elucidated. Hence also candidate genes were analysed, which could not be assigned to the above listed categories. The present invention is based at least in part on the discovery of polymorphisms, that lie in genomic regions of unknown physiological function.

Results

After conducting an association study, we surprisingly found polymorphic sites in a number of candidate genes which show a strong correlation with the following phenotypes of the patients analysed: "Healthy" as used herein refers to individuals that neither suffer from existing CVD, nor exhibit an increased risk for CVD through their serum lipid level profile. "CVD prone" as used herein refers to individuals with existing CVD and/or a serum lipid profile that confers a high risk to get CVD (see Table 1a for definitions of healthy and CVD prone serum lipid levels). "High responder" as used herein refers to patients who benefit from relatively small amounts of a given drug. "Low responder" as used herein refers to patients who need relatively high doses in order to obtain benefit from the medication. "Tolerant patient" refers to individuals who can tolerate high doses of a medicament without exhibiting adverse drug reactions. "ADR patient" as used herein refers to individuals who suffer from ADR or show clinical symptoms (like creatine kinase elevation in

blood) even after receiving only minor doses of a medicament (see Table 1b for a detailed definition of drug response phenotypes).

5 Polymorphic sites in candidate genes that were found to be significantly associated with either of the above mentioned phenotypes will be referred to as "phenotype associated SNPs" (PA SNPs). The respective genomic loci that harbour PA SNPs will be referred to as "phenotype associated genes" (PA genes), irrespective of the actual function of this gene locus.

10 In particular we surprisingly found PA SNPs associated with CVD, drug efficacy, (EFF) or adverse drug reactions (ADR) in the following genes.

ABCB11: ATP-binding cassette, sub-family B (MDR/TAP), member 11

15 The membrane-associated protein encoded by this gene is a member of the superfamily of ATP-binding cassette (ABC) transporters. ABC proteins transport various molecules across extra- and intra-cellular membranes. ABC genes are divided into seven distinct subfamilies (ABC1, MDR/TAP, MRP, ALD, OABP, GCN20, White). This protein is a member of the MDR/TAP subfamily. Members of

20 the MDR/TAP subfamily are involved in multidrug resistance. The protein encoded by this gene is the major canalicular bile salt export pump in man. Mutations in this gene cause a form of progressive familial intrahepatic cholestases which are a group of inherited disorders with severe cholestatic liver disease from early infancy.

ABCB4: ATP-binding cassette, sub-family B (MDR/TAP), member 4

The membrane-associated protein encoded by this gene is a member of the superfamily of ATP-binding cassette (ABC) transporters. ABC proteins transport various molecules across extra- and intra-cellular membranes. ABC genes are

30 divided into seven distinct subfamilies (ABC1, MDR/TAP, MRP, ALD, OABP, GCN20, White). This protein is a member of the MDR/TAP subfamily. Members of

the MDR/TAP subfamily are involved in multidrug resistance as well as antigen presentation. This gene encodes a full transporter and member of the p-glycoprotein family of membrane proteins with phosphatidylcholine as its substrate. The function of this protein has not yet been determined; however, it may involve transport of phospholipids from liver hepatocytes into bile. Alternative splicing of this gene results in several products of undetermined function.

ABCC1: ATP-binding cassette, sub-family C (CFTR/MRP), member 1

The protein encoded by this gene is a member of the superfamily of ATP-binding cassette (ABC) transporters. ABC proteins transport various molecules across extra- and intra-cellular membranes. ABC genes are divided into seven distinct subfamilies (ABC1, MDR/TAP, MRP, ALD, OABP, GCN20, White). This full transporter is a member of the MRP subfamily which is involved in multi-drug resistance. This protein functions as a multispecific organic anion transporter, with oxidized glutathione, cysteinyl leukotrienes, and activated aflatoxin B1 as substrates. This protein also transports glucuronides and sulfate conjugates of steroid hormones and bile salts. Alternative splicing by exon deletion results in several splice variants but maintains the original open reading frame in all forms.

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ACTB mRNA for mutant beta-actin

Beta actin is one of six different actin isoforms which have been identified. ACTB is one of the two nonmuscle cytoskeletal actins. Actins are highly conserved proteins that are involved in cell motility, structure and integrity. Alpha actins are a major constituent of the contractile apparatus.

ACTIN, ALPHA SKELETAL MUSCLE (ALPHA-ACTIN 1).

Actin alpha 1 which is expressed in skeletal muscle is one of six different actin isoforms which have been identified. Actins are highly conserved proteins that are

involved in cell motility, structure and integrity. Alpha actins are a major constituent of the contractile apparatus.

ADCYAP1: adenylate cyclase activating polypeptide 1 (pituitary)

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This gene encodes adenylate cyclase activating polypeptide 1. Mediated by adenylate cyclase activating polypeptide 1 receptors, this polypeptide stimulates adenylate cyclase and subsequently increases the cAMP level in target cells. Adenylate cyclase activating polypeptide 1 is not only a hypophysiotropic hormone, but also functions 10 as a neurotransmitter and neuromodulator. In addition, it plays a role in paracrine and autocrine regulation of certain types of cells. This gene is composed of five exons. Exons 1 and 2 encode the 5' UTR and signal peptide, respectively; exon 4 encodes an adenylate cyclase activating polypeptide 1-related peptide; and exon 5 encodes the mature peptide and 3' UTR. This gene encodes three different mature peptides, 15 including two isotypes: a shorter form and a longer form.

ADRB3: adrenergic, beta-3-, receptor

The ADRB3 gene product, beta-3-adrenergic receptor, is located mainly in adipose 20 tissue and is involved in the regulation of lipolysis and thermogenesis. Beta adrenergic receptors are involved in the epinephrine and norepinephrine-induced activation of adenylate cyclase through the action of G proteins.

AGL: amylo-1, 6-glucosidase, 4-alpha-glucanotransferase (glycogen debranching enzyme, glycogen storage disease type III)

Glycogen debranching enzyme is involved in glycogen degradation and has two independent catalytic activities: a 4-alpha-glucotransferase activity (EC 2.4.1.25) and a amylo-1,6-glucosidase activity (EC 3.4.1.33). Both activities occur at different sites 30 on the single polypeptide chain. Mutations in this gene cause glycogen storage disease. A wide range of clinical and enzymatic variability occurs in glycogen

debrancher deficiency, some of which may be due to tissue-specific alternative splicing. Six splice variants that differ in the 5' end have been identified in liver and muscle tissue. Variants 1, 5, and 6 are present in both liver and muscle, whereas variants 2, 3, and 4 occur in muscle. Variants 1 through 4 encode identical proteins (isoform 1) that include 27 N-terminal amino acids not found in splice variants 5 and 6. Variants 5 and 6 encode different amino-terminal ends of 10 and 11 amino acids in protein isoforms 2 and 3, respectively, with the remainder of the peptide identical to that of isoforms 1..

10 **AKAP1: A kinase (PRKA) anchor protein 1**

Anchors cAMP-dependent protein kinase near its physiological substrates, interacts with both the type I and type II regulatory subunits.

15 **Angiotensinogen gene**

The protein encoded by this gene, pre-angiotensinogen or angiotensinogen precursor, is expressed in the liver and is cleaved by the enzyme renin in response to lowered blood pressure. The resulting product, angiotensin I is then cleaved by angiotensin converting enzyme (ACE) to generate the physiologically active enzyme angiotensin II. The protein is involved in maintaining blood pressure and in the pathogenesis of essential hypertension and preeclampsia.

25 **ANXA6: annexin A6**

Annexin VI belongs to a family of calcium-dependent membrane and phospholipid binding proteins. Although their functions are still not clearly defined, several members of the annexin family have been implicated in membrane-related events along exocytotic and endocytotic pathways. The annexin VI gene is approximately 60 kbp long and contains 26 exons. It encodes a protein of about 68 kDa that consists of eight 68-amino acid repeats separated by linking sequences of variable lengths. It

is highly similar to human annexins I and II sequences, each of which contain four such repeats. Exon 21 of annexin VI is alternatively spliced, giving rise to two isoforms that differ by a 6-amino acid insertion at the start of the seventh repeat. Annexin VI has been implicated in mediating the endosome aggregation and vesicle fusion in secreting epithelia during exocytosis.

AP2B1: adaptor-related protein complex 2, beta 1 subunit

The beta adaptin subunit is part of the clathrin coat assembly complex which links clathrin to receptors in coated pits and vesicles. These vesicles are involved in endocytosis and Golgi processing. The beta 1 subunit is one of the assembly proteins which binds to clathrin and initiates coat formation.

APOA1: apolipoprotein A-I

APOA1 promotes cholesterol efflux from tissues to the liver for excretion. Apolipoprotein A-I is the major protein component of high density lipoprotein (HDL) in the plasma. Synthesized in the liver and small intestine, it consists of two identical chains of 77 amino acids; an 18-amino acid signal peptide is removed co-translationally and a 6-amino acid propeptide is cleaved post-translationally. Variation in the latter step, in addition to modifications leading to so-called isoforms, is responsible for some of the polymorphism observed. APOA1 is a cofactor for lecithin cholesterol acyltransferase (LCAT) which is responsible for the formation of most plasma cholesteryl esters. The APOA1, APOC3 and APOA4 genes are closely linked in both rat and human genomes. The A-I and A-IV genes are transcribed from the same strand, while the C-III gene is transcribed convergently in relation to A-I. Defects in the apolipoprotein A-1 gene are associated with HDL deficiency and Tangier disease.

APOA4: ap lip protein A-IV

Apolipoprotein (apo) A-IV gene contains 3 exons separated by two introns. A sequence polymorphism has been identified in the 3'UTR of the third exon. The primary translation product is a 396-residue preprotein which after proteolytic processing is secreted its primary site of synthesis, the intestine, in association with chylomicron particles. Although its precise function is not known, apo A-IV is a potent activator of lecithin-cholesterol acyltransferase in vitro.

10 APOB: apolipoprotein B

Apolipoprotein B (ApoB) is the main apolipoprotein of chylomicrons and low density lipoproteins (LDL). The protein occurs in the plasma in 2 main isoforms, apoB-48 and apoB-100. The first is synthesized exclusively by the gut, the second by the liver. The intestinal (B-48) and hepatic (B-100) forms of apoB are coded by a single gene and by a single mRNA transcript larger than 16 kb. The 2 proteins share a common amino terminal sequence. In the ApoB-100 isoform the precursor has 4,563 amino acids, and the mature apoB-100 has 4,536 amino acid residues. Mature, circulating B-48 is homologous over its entire length (estimated to be between 2,130 and 2,144 amino acid residues) with the amino-terminal portion of B-100 and contains no sequence from the carboxyl end of B-100. From structural studies, it is thought that apoB-48 represents the amino-terminal 47% of apoB-100 and that the carboxyl terminus of apoB-48 is in the vicinity of residue 2151 of apoB-100. Apolipoprotein B-48 may be the product of an intestinal mRNA with an in-frame UAA stop codon resulting from a C-to-U change in the codon CAA encoding Gln(2153) in apoB-100 mRNA. Since only the sequence that codes B-100 is present in genomic DNA, this presents the possibility of an organ-specific introduction of a stop codon to an mRNA and the change from CAA to UAA of codon 2153 of the message as a unique RNA editing process..

APOD: apolipoprotein D

Apolipoprotein D (Apo-D) is a component of high density lipoprotein that has no marked similarity to other apolipoprotein sequences. It has a high degree of homology to plasma retinol-binding protein and other members of the alpha 2 microglobulin protein superfamily of carrier proteins, also known as lipocalins. It is a glycoprotein of estimated molecular weight 33 KDa. Apo-D is closely associated with the enzyme lecithin:cholesterol acyltransferase - an enzyme involved in lipoprotein metabolism.

Apolipoprotein B

Apolipoprotein B (ApoB) is the main apolipoprotein of chylomicrons and low density lipoproteins (LDL). The protein occurs in the plasma in 2 main isoforms, apoB-48 and apoB-100. The first is synthesized exclusively by the gut, the second by the liver. The intestinal (B-48) and hepatic (B-100) forms of apoB are coded by a single gene and by a single mRNA transcript larger than 16 kb. The 2 proteins share a common amino terminal sequence. In the ApoB-100 isoform the precursor has 4,563 amino acids, and the mature apoB-100 has 4,536 amino acid residues. Mature, circulating B-48 is homologous over its entire length (estimated to be between 2,130 and 2,144 amino acid residues) with the amino-terminal portion of B-100 and contains no sequence from the carboxyl end of B-100. From structural studies, it is thought that apoB-48 represents the amino-terminal 47% of apoB-100 and that the carboxyl terminus of apoB-48 is in the vicinity of residue 2151 of apoB-100. Apolipoprotein B-48 may be the product of an intestinal mRNA with an in-frame UAA stop codon resulting from a C-to-U change in the codon CAA encoding Gln(2153) in apoB-100 mRNA. Since only the sequence that codes B-100 is present in genomic DNA, this presents the possibility of an organ-specific introduction of a stop codon to an mRNA and the change from CAA to UAA of codon 2153 of the message as a unique RNA editing process..

APXL: apical protein-like (Xenopus laevis)

The protein encoded by this gene shares significant similarities with the apical protein from *Xenopus laevis* which is implicated in amiloride-sensitive sodium channel activity. This gene is a strong candidate gene for ocular albinism type 1 syndrome.

ARF4: ADP-ribosylation factor 4

10 ADP-ribosylation factor 4 (ARF4) is a member of the human ARF gene family. These genes encode small guanine nucleotide-binding proteins that stimulate the ADP-ribosyltransferase activity of cholera toxin and play a role in vesicular trafficking and as activators of phospholipase D. The gene products include 6 ARF proteins and 11 ARF-like proteins and constitute 1 family of the RAS superfamily.

15 The ARF proteins are categorized as class I (ARF1, ARF2, and ARF3), class II (ARF4 and ARF5) and class III (ARF6). The members of each class share a common gene organization. The ARF4 gene spans approximately 12kb and contains six exons and five introns. The ARF4 is the most divergent member of the human ARFs. Conflicting Map positions at 3p14 or 3p21 have been reported for this gene.

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ATP1A2: ATPase, Na⁺/K⁺ transporting, alpha 2 (+) polypeptide

Alpha 2 subunit of the sodium- and potassium-transporting ATPase; required for Na⁺ and K⁺ gradient maintenance across plasma membrane.

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ATP1B1: ATPase, Na⁺/K⁺ transporting, beta 1 polypeptide

Beta 1 subunit of Na⁺/K⁺-ATPase.

ATP1B3: ATPase, Na⁺/K⁺ transporting, beta 3 polypeptide

30 Beta 3 subunit of the Na⁺/K⁺-ATPase.

ATP2A2: ATPase, Ca++ transporting, cardiac muscle, slow twitch 2

Slow twitch cardiac muscle Ca²⁺-ATPase; pumps calcium, may have a role in calcium signaling pathways.

- 5 **ATP5G1: ATP synthase, H⁺ transporting, mitochondrial F0 complex, subunit c (subunit 9), isoform 1**

Isoform 1 (P1) of subunit c, H⁺-translocating subunit of F0 ATP synthase; catalyzes the synthesis of ATP during oxidative phosphorylation.

- 10 **ATP6V1E: ATPase, H⁺ transporting, lysosomal 31kD, V1 subunit E**

This gene encodes a component of vacuolar ATPase (V-ATPase), a multisubunit enzyme that mediates acidification of eukaryotic intracellular organelles. V-ATPase dependent organelle acidification is necessary for such intracellular processes as protein sorting, zymogen activation, and receptor-mediated endocytosis. V-ATPase is comprised of a cytosolic V1 domain and a transmembrane V0 domain. The V1 domain consists of a hexamer of three A and three B subunits plus the C, D, and E subunits. It contains the ATP catalytic site. The encoded protein is known as the E subunit and is found ubiquitously. Pseudogenes for this gene have been found in the genome.

ATPase, Ca++ transporting, cardiac muscle, fast twitch 1

- 25 Fast-twitch skeletal muscle sarcoplasmic reticulum Ca²⁺-ATPase; pumps calcium.

AXIN1: axin

Strongly similar to murine Axin; may regulate embryonic axis formation.

BMPR1A: bone morphogenetic protein receptor, type IA

The bone morphogenetic protein (BMP) receptors are a family of transmembrane serine/threonine kinases that include the type I receptors BMPR1A and BMPR1B and the type II receptor BMPR2. These receptors are also closely related to the activin receptors, ACVR1 and ACVR2. The ligands of these receptors are members of the TGF-beta superfamily. TGF-betas and activins transduce their signals through the formation of heteromeric complexes with 2 different types of serine (threonine) kinase receptors: type I receptors of about 50-55 kD and type II receptors of about 70-80 kD. Type II receptors bind ligands in the absence of type I receptors, but they require their respective type I receptors for signaling, whereas type I receptors require their respective type II receptors for ligand binding.

BRD3: bromodomain containing 3

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This gene was identified based on its homology to the gene encoding the RING3 protein, a serine/threonine kinase. The gene localizes to 9q34, a region which contains several major histocompatibility complex (MHC) genes. The function of the encoded protein is not known.

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CACNA1C: calcium channel, voltage-dependent, L type, alpha 1C subunit

Alpha 1C subunit of the voltage-dependent calcium channel; channel is of the L type and is expressed in the heart.

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CALB2: calbindin 2, (29kD, calretinin)

Calbindin 2 (calretinin), closely related to calbindin 1, is an intracellular calcium-binding protein belonging to the troponin C superfamily. Calbindin 1 is known to be involved in the vitamin-D-dependent calcium absorption through intestinal and renal epithelia, while the function of neuronal calbindin 1 and calbindin 2 is poorly

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understood. The sequence of the calbindin 2 cDNA reveals an open reading frame of 271 codons coding for a protein of 31,520 Da, and shares 58% identical residues with human calbindin 1. Calbindin 2 contains five presumably active and one presumably inactive calcium-binding domains. Comparison with the partial sequences available for chick and guinea pig calbindin 2 reveals that the protein is highly conserved in evolution. The calbindin 2 message was detected in the brain, while absent from heart muscle, kidney, liver, lung, spleen, stomach and thyroid gland. There are two additional forms of alternatively spliced calbindin 2 mRNAs encoding C-terminally truncated proteins. Exon 7 can splice to exon 9, resulting in a frame shift and a translational stop at the second codon of exon 9, and encoding calretinin-20k. Exon 7 can also splice to exon 10, resulting in a frame shift and a translational stop at codon 15 of exon 10, and encoding calretinin-22k. The truncated proteins are able to bind calcium.

15 **CALCIUM-TRANSPORTING ATPASE PLASMA MEMBRANE, ISOFORMS
3A/3B (EC 3.6.1.38) (CALCIUM PUMP) (PMCA3).**

Plasma membrane Ca²⁺-ATPase 3; pumps calcium.

20 **CALM3: calmodulin 3 (phosphorylase kinase, delta)**

Calmodulin 3; binds calcium.

CAV1: caveolin 1, caveolae protein, 22kD

25 The scaffolding protein encoded by this gene is the main component of the caveolae plasma membranes found in most cell types. The protein links integrin subunits to the tyrosine kinase FYN, an initiating step in coupling integrins to the Ras-ERK pathway and promoting cell cycle progression. The gene is a tumor suppressor gene candidate and a negative regulator of the Ras-p42/44 MAP kinase cascade. CAV1 and CAV2 are located next to each other on chromosome 7 and express colocalizing

proteins that form a stable hetero-oligomeric complex. By using alternative initiation codons in the same reading frame, two isoforms (alpha and beta) are encoded by a single transcript from this gene.

5 **CAV3: caveolin 3**

This gene encodes a caveolin family member, which functions as a component of the caveolae plasma membranes found in most cell types. Caveolin proteins are proposed to be scaffolding proteins for organizing and concentrating certain caveolin-interacting molecules. Mutations identified in this gene lead to interference with protein oligomerization or intra-cellular routing, disrupting caveolae formation and resulting in Limb-Girdle muscular dystrophy type-1C (LGMD-1C), hyperCKemia or rippling muscle disease (RMD). Alternative splicing has been identified for this locus, with inclusion or exclusion of a differentially spliced intron. In addition, transcripts utilize multiple polyA sites and contain two potential translation initiation sites.

CCR2: chemokine (C-C motif) receptor 2

20 This gene encodes two isoforms of a receptor for monocyte chemoattractant protein-1, a chemokine which specifically mediates monocyte chemotaxis. Monocyte chemoattractant protein-1 is involved in monocyte infiltration in inflammatory diseases such as rheumatoid arthritis as well as in the inflammatory response against tumors. The receptors encoded by this gene mediate agonist-dependent calcium mobilization and inhibition of adenylyl cyclase. This gene is located in the chemokine receptor gene cluster region. Two alternatively spliced transcript variants are expressed by the gene.

CDH1: cadherin 1, type 1, E-cadherin (epithelial)

This gene is a classical cadherin from the cadherin superfamily. The encoded protein is a calcium dependent cell-cell adhesion glycoprotein comprised of five extracellular cadherin repeats, a transmembrane region and a highly conserved cytoplasmic tail.

5 Mutations in this gene are correlated with gastric, breast, colorectal, thyroid and ovarian cancer. Loss of function is thought to contribute to progression in cancer by increasing proliferation, invasion, and/or metastasis. The ectodomain of this protein mediates bacterial adhesion to mammalian cells and the cytoplasmic domain is required for internalization. Identified transcript variants arise from mutation at 10 consensus splice sites.

CDH11: cadherin 11, type 2, OB-cadherin (osteoblast)

15 This gene encodes a type II classical cadherin from the cadherin superfamily, integral membrane proteins that mediate calcium-dependent cell-cell adhesion. Mature cadherin proteins are composed of a large N-terminal extracellular domain, a single membrane-spanning domain, and a small, highly conserved C-terminal cytoplasmic domain. Type II (atypical) cadherins are defined based on their lack of a HAV cell adhesion recognition sequence specific to type I cadherins. Expression of this 20 particular cadherin in osteoblastic cell lines, and its upregulation during differentiation, suggests a specific function in bone development and maintenance. Two splice variants have been identified, one of which encodes an isoform with a truncated cytoplasmic domain.

CDH13: cadherin 13, H-cadherin (heart)

This gene is a member of the cadherin superfamily. The encoded protein is a calcium dependent cell-cell adhesion glycoprotein comprised of five extracellular cadherin repeats, a transmembrane region but, unlike the typical cadherin superfamily member, lacks the highly conserved cytoplasmic region. This particular cadherin is a

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putative mediator of cell-cell interaction in the heart and may act as a negative regulator of neural cell growth. The gene locus is hypermethylated or deleted in breast, ovarian and lung cancers. Two major mRNA transcripts encoding identical proteins are found, products of alternative polyadenylation sites.

5

CENPC1: centromere protein C 1

Centromere protein C 1 is a centromere autoantigen and a component of the inner kinetochore plate. The protein is required for maintaining proper kinetochore size
10 and a timely transition to anaphase. A putative pseudogene exists on chromosome 12.

Cholesteryl ester transfer protein (CETP)

Cholesteryl ester transfer protein (CETP) transfers cholesteryl esters between
15 lipoproteins. CETP may effect susceptibility to atherosclerosis.

CLCN4: chloride channel 4

The CLCN family of voltage-dependent chloride channel genes comprises nine
20 members (CLCN1-7, Ka and Kb) which demonstrate quite diverse functional characteristics while sharing significant sequence homology. Chloride channel 4 has an evolutionary conserved CpG island and is conserved in both mouse and hamster. This gene is mapped in close proximity to APXL (Apical protein Xenopus laevis-like) and OA1 (Ocular albinism type I), which are both located on the human X
25 chromosome at band p22.3. The physiological role of chloride channel 4 remains unknown but may contribute to the pathogenesis of neuronal disorders.

CLCNKA: chloride channel Ka

30 Putative chloride channel; member of the CLC family of voltage-gated chloride channels.

COL6A3: collagen, type VI, alpha 3

This gene encodes the alpha 3 chain, one of the three alpha chains of type VI collagen, a beaded filament collagen found in most connective tissues. The alpha 3 chain of type VI collagen is much larger than the alpha 1 and 2 chains. This difference in size is largely due to an increase in the number of subdomains, similar to von Willebrand Factor type A domains, found in the amino terminal globular domain of all the alpha chains. These domains have been shown to bind extracellular matrix proteins, an interaction that explains the importance of this collagen in organizing matrix components. Mutations in the type VI collagen genes are associated with Bethlem myopathy. In addition to the full length transcript, four transcript variants have been identified that encode proteins with N-terminal globular domains of varying sizes.

COL7A1: collagen, type VII, alpha 1 (epidermolysis bullosa, dystrophic, dominant and recessive)

This gene encodes the alpha chain of type VII collagen. The type VII collagen fibril, composed of three identical alpha collagen chains, is restricted to the basement zone beneath stratified squamous epithelia. It functions as an anchoring fibril between the external epithelia and the underlying stroma. Mutations in this gene are associated with all forms of dystrophic epidermolysis bullosa. In the absence of mutations, however, an acquired form of this disease can result from an autoimmune response made to type VII collagen.

COL9A3: collagen, type IX, alpha 3

This gene encodes one of the three alpha chains of type IX collagen, the major collagen component of hyaline cartilage. Type IX collagen, a heterotrimeric

molecule, is usually found in tissues containing type II collagen, a fibrillar collagen. Mutations in this gene are associated with multiple epiphyseal dysplasia.

COMT: catechol-O-methyltransferase

5

Catechol-O-methyltransferase catalyzes the transfer of a methyl group from S-adenosylmethionine to catecholamines, including the neurotransmitters dopamine, epinephrine, and norepinephrine. This O-methylation results in one of the major degradative pathways of the catecholamine transmitters. In addition to its role in the metabolism of endogenous substances, COMT is important in the metabolism of catechol drugs used in the treatment of hypertension, asthma, and Parkinson disease. COMT is found in two forms in tissues, a soluble form (S-COMT) and a membrane-bound form (MB-COMT). The differences between S-COMT and MB-COMT reside within the N-termini. The transcript variants are formed through the use of alternative translation initiation sites and promoters.

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COX10: COX10 homolog, cytochrome c oxidase assembly protein, heme A:farnesyltransferase (yeast)

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Cytochrome c oxidase (COX), the terminal component of the mitochondrial respiratory chain, catalyzes the electron transfer from reduced cytochrome c to oxygen. This component is a heteromeric complex consisting of 3 catalytic subunits encoded by mitochondrial genes and multiple structural subunits encoded by nuclear genes. The mitochondrially-encoded subunits function in electron transfer, and the nuclear-encoded subunits may function in the regulation and assembly of the complex. This nuclear gene encodes heme A:farnesyltransferase, which is not a structural subunit but required for the expression of functional COX and functions in the maturation of the heme A prosthetic group of COX. This protein is predicted to contain 7-9 transmembrane domains localized in the mitochondrial inner membrane. A gene mutation, which results in the substitution of a lysine for an asparagine (N204K), is identified to be responsible for cytochrome c oxidase deficiency. In

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addition, this gene is disrupted in patients with CMT1A (Charcot-Marie-Tooth type 1A) duplication and with HNPP (hereditary neuropathy with liability to pressure palsies) deletion.

5 **CPB2: carboxypeptidase B2 (plasma, carboxypeptidase U)**

Carboxypeptidases are enzymes that hydrolyze C-terminal peptide bonds. The carboxypeptidase family includes metallo-, serine, and cysteine carboxypeptidases. According to their substrate specificity, these enzymes are referred to as carboxypeptidase A (cleaving aliphatic residues) or carboxypeptidase B (cleaving basic amino residues). The protein encoded by this gene is activated by trypsin and acts on carboxypeptidase B substrates. After thrombin activation, the mature protein downregulates fibrinolysis. Polymorphisms have been described for this gene and its promoter region. Available sequence data analyses indicate splice variants that encode different isoforms.

CPO: coproporphyrinogen oxidase (coproporphyrin, harderoporphyrin)

20 Coproporphyrinogen; catalyzes oxidative decarboxylation in sixth step of heme biosynthesis.

CRYAB: crystallin, alpha B

Crystallins are separated into two classes: taxon-specific, or enzyme, and ubiquitous. 25 The latter class constitutes the major proteins of vertebrate eye lens and maintains the transparency and refractive index of the lens. Since lens central fiber cells lose their nuclei during development, these crystallins are made and then retained throughout life, making them extremely stable proteins. Mammalian lens crystallins are divided into alpha, beta, and gamma families; beta and gamma crystallins are also considered as a superfamily. Alpha and beta families are further divided into acidic and basic groups. Seven protein regions exist in crystallins: four homologous motifs, a

connecting peptide, and N- and C-terminal extensions. Alpha crystallins are composed of two gene products: alpha-A and alpha-B, for acidic and basic, respectively. Alpha crystallins can be induced by heat shock and are members of the small heat shock protein (sHSP also known as the HSP20) family. They act as molecular chaperones although they do not renature proteins and release them in the fashion of a true chaperone; instead they hold them in large soluble aggregates. Post-translational modifications decrease the ability to chaperone. These heterogeneous aggregates consist of 30-40 subunits; the alpha-A and alpha-B subunits have a 3:1 ratio, respectively. Two additional functions of alpha crystallins are an autokinase activity and participation in the intracellular architecture. Alpha-A and alpha-B gene products are differentially expressed; alpha-A is preferentially restricted to the lens and alpha-B is expressed widely in many tissues and organs. Elevated expression of alpha-B crystallin occurs in many neurological diseases; a missense mutation cosegregated in a family with a desmin-related myopathy..

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CSF2RB: colony stimulating factor 2 receptor, beta, low-affinity (granulocyte-macrophage)

20 CSF2RB is a common beta chain of the high affinity receptor for IL-3, IL-5 and CSF. Defective CSF2RB has been reported to be associated with protein alveolar proteinosis.

CUBN: cubilin (intrinsic factor-cobalamin receptor)

25 Cubilin (CUBN) acts as a receptor for intrinsic factor-vitamin B12 complexes. The role of receptor is supported by the presence of 27 CUB domains. Cubilin is located within the epithelium of intestine and kidney. Mutations in CUBN may play a role in autosomal recessive megaloblastic anemia.

30 **CXorf6: chromosome X open reading frame 6**

CYP17: cytochrome P450, subfamily XVII (steroid 17-alpha-hydroxylase), adrenal hyperplasia

This gene encodes a member of the cytochrome P450 superfamily of enzymes. The

5 cytochrome P450 proteins are monooxygenases which catalyze many reactions involved in drug metabolism and synthesis of cholesterol, steroids and other lipids.

This protein localizes to the endoplasmic reticulum. It has both 17alpha-hydroxylase and 17,20-lyase activities and is a key enzyme in the steroidogenic pathway that produces progestins, mineralocorticoids, glucocorticoids, androgens, and estrogens.

10 Mutations in this gene are associated with isolated steroid-17 alpha-hydroxylase deficiency, 17-alpha-hydroxylase/17,20-lyase deficiency, pseudohermaphroditism, and adrenal hyperplasia.

**CYP2C8: cytochrome P450, subfamily IIIC (mephenytoin 4-hydroxylase),
15 polypeptide 8**

This gene encodes a member of the cytochrome P450 superfamily of enzymes. The cytochrome P450 proteins are monooxygenases which catalyze many reactions involved in drug metabolism and synthesis of cholesterol, steroids and other lipids.

20 This protein localizes to the endoplasmic reticulum and its expression is induced by phenobarbital. The enzyme is known to metabolize many xenobiotics, including the anticonvulsive drug mephenytoin, benzo(a)pyrene, 7-ethoxycoumarin, and the anti-cancer drug taxol. Two transcript variants for this gene have been described; it is thought that the longer form does not encode an active cytochrome P450 since its protein product lacks the heme binding site. This gene is located within a cluster of 25 cytochrome P450 genes on chromosome 10q24.

CYP2E: cytochrome P450, subfamily IIIE (ethanol-inducible)

30 This gene encodes a member of the cytochrome P450 superfamily of enzymes. The cytochrome P450 proteins are monooxygenases which catalyze many reactions

involved in drug metabolism and synthesis of cholesterol, steroids and other lipids. This protein localizes to the endoplasmic reticulum and is induced by ethanol, the diabetic state, and starvation. The enzyme metabolizes both endogenous substrates, such as ethanol, acetone, and acetal, as well as exogenous substrates including benzene, carbon tetrachloride, ethylene glycol, and nitrosamines which are premutagens found in cigarette smoke. Due to its many substrates, this enzyme may be involved in such varied processes as gluconeogenesis, hepatic cirrhosis, diabetes, and cancer.

10 **CYP3A4**

This gene, CYP3A4, encodes a member of the cytochrome P450 superfamily of enzymes. The cytochrome P450 proteins are monooxygenases which catalyze many reactions involved in drug metabolism and synthesis of cholesterol, steroids and other lipids. This protein localizes to the endoplasmic reticulum and its expression is induced by glucocorticoids and some pharmacological agents. This enzyme is involved in the metabolism of approximately half the drugs which are used today, including acetaminophen, codeine, cyclosporin A, diazepam and erythromycin. The enzyme also metabolizes some steroids and carcinogens. This gene is part of a cluster of cytochrome P450 genes on chromosome 7q21.1. Previously another CYP3A gene, CYP3A3, was thought to exist; however, it is now thought that this sequence represents a transcript variant of CYP3A4.

25 **CYP4F8: cytochrome P450, subfamily IVF, polypeptide 8**

This gene, CYP4F8, encodes a member of the cytochrome P450 superfamily of enzymes. The cytochrome P450 proteins are monooxygenases which catalyze many reactions involved in drug metabolism and synthesis of cholesterol, steroids and other lipids. This protein localizes to the endoplasmic reticulum and functions as a 19-hydroxylase of prostaglandins in seminal vesicles. This gene is part of a cluster of

cytochrome P450 genes on chromosome 19. Another member of this family, CYP4F3, is approximately 18 kb away.

5 CYP8B1: cytochrome P450, subfamily VIIIB (sterol 12-alpha-hydroxylase), polypeptide 1

This gene encodes a member of the cytochrome P450 superfamily of enzymes. The cytochrome P450 proteins are monooxygenases which catalyze many reactions involved in drug metabolism and synthesis of cholesterol, steroids and other lipids.

10 This endoplasmic reticulum membrane protein catalyzes the conversion of 7 alpha-hydroxy-4-cholesten-3-one into 7-alpha,12-alpha-dihydroxy-4-cholesten-3-one. The balance between these two steroids determines the relative amounts of cholic acid and chenodeoxycholic acid both of which are secreted in the bile and affect the solubility of cholesterol. This gene is unique among the cytochrome P450 genes in

15 that it is intronless.

DBI: diazepam binding inhibitor (GABA receptor modulator, acyl-Coenzyme A binding protein)

20 Diazepam binding inhibitor (acyl-CoA-binding protein); binds and induces medium-chain acyl-CoA ester synthesis.

DEF A6: defensin, alpha 6, Paneth cell-specific

25 Defensins are a family of microbicidal and cytotoxic peptides thought to be involved in host defense. They are abundant in the granules of neutrophils and also found in the epithelia of mucosal surfaces such as those of the intestine, respiratory tract, urinary tract, and vagina. Members of the defensin family are highly similar in protein sequence and distinguished by a conserved cysteine motif. Several alpha defensin genes appear to be clustered on chromosome 8. The protein encoded by this

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gene, defensin, alpha 6, is highly expressed in the secretory granules of Paneth cells of the small intestine, and likely plays a role in host defense of human bowel.

DEK: DEK oncogene (DNA binding)

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Site-specific DNA binding protein; involved in transcriptional regulation and signal transduction.

DFNA5: deafness, autosomal dominant 5

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Hearing impairment is a heterogeneous condition with over 40 loci described. The protein encoded by this gene is expressed in fetal cochlea, however, its function is not known. Nonsyndromic hearing impairment is associated with a mutation in this gene.

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DGKD: diacylglycerol kinase, delta (130kD)

Diacylglycerol kinase delta; phosphorylates the arachidonoyl type of diacylglycerol; contains a pleckstrin homology domain and an EPH domain.

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DOCK1: dedicator of cyto-kinesis 1

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Dedicator of cyto-kinesis 1 binds to the SH3 domain of CRK protein. It may regulate cell surface extension and may have a role in the cell surface extension of an engulfing cell around a dying cell during apoptosis.

ECE1: endothelin converting enzyme 1

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Endothelin converting enzyme; metalloprotease that regulates a peptide involved in vasoconstriction.

E-Selectin (CD62E)

The endothelial leukocyte adhesion molecule-1 is expressed by cytokine-stimulated endothelial cells. It is thought to be responsible for the accumulation of blood leukocytes at sites of inflammation by mediating the adhesion of cells to the vascular lining. It exhibits structural features such as the presence of lectin- and EGF-like domains followed by short consensus repeat (SCR) domains that contain 6 conserved cysteine residues. These proteins are part of the selectin family of cell adhesion molecules. This gene is present in single copy in the human genome and contains 14 exons spanning about 13 kb of DNA. Adhesion molecules participate in the interaction between leukocytes and the endothelium and appear to be involved in the pathogenesis of atherosclerosis.

ESR1: estrogen receptor 1

15 Estrogen receptor; nuclear receptor transcription factor activated by ligand-binding, involved in hormone-mediated inhibition of gene expression.

ESR2: estrogen receptor 2 (ER beta)

20 Estrogen receptor beta 2; transcriptional activator involved in regulation of reproduction; exists in five isoforms.

F2: coagulation factor II (thrombin)

25 Coagulation factor II is proteolytically cleaved to form thrombin in the first step of the coagulation cascade which ultimately results in the stemming of blood loss. F2 also plays a role in maintaining vascular integrity during development and postnatal life. Mutations in F2 leads to various forms of thrombosis and dysprothrombinemia.

F3: coagulation factor III (thromboplastin, tissue factor)

This gene encodes coagulation factor III which is a cell surface glycoprotein. This factor enables cells to initiate the blood coagulation cascades, and it functions as the high-affinity receptor for the coagulation factor VII. The resulting complex provides a catalytic event that is responsible for initiation of the coagulation protease cascades by specific limited proteolysis. Unlike the other cofactors of these protease cascades, which circulate as nonfunctional precursors, this factor is a potent initiator that is fully functional when expressed on cell surfaces. There are 3 distinct domains of this factor: extracellular, transmembrane, and cytoplasmic. This protein is the only one in the coagulation pathway for which a congenital deficiency has not been described.

F5: coagulation factor V (proaccelerin, labile factor)

This gene encodes coagulation factor V which is an essential factor of the blood coagulation cascade. This factor circulates in plasma, and is converted to the active form by the release of the activation peptide by thrombin during coagulation. This generates a heavy chain and a light chain which are held together by calcium ions. The active factor V is a cofactor that participates with activated coagulation factor X to activate prothrombin to thrombin. Defects in this gene result in either an autosomal recessive hemorrhagic diathesis or an autosomal dominant form of thrombophilia, which is known as activated protein C resistance.

F7: coagulation factor VII (serum prothrombin conversion accelerator)

This gene encodes coagulation factor VII which is a vitamin K-dependent factor essential for hemostasis. This factor circulates in the blood in a zymogen form, and is converted to an active form by either factor IXa, factor Xa, factor XIIa, or thrombin by minor proteolysis. Upon activation of the factor VII, a heavy chain containing a catalytic domain and a light chain containing 2 EGF-like domains are generated, and two chains are held together by a disulfide bond. In the presence of factor III and

calcium ions, the activated factor then further activates the coagulation cascade by converting factor IX to factor IXa and/or factor X to factor Xa. Alternative splicing of this gene results in 2 transcripts. Defects in this gene can cause coagulopathy.

5 **F9: coagulation factor IX (plasma thromboplastic component, Christmas disease, hemophilia B)**

This gene encodes vitamin K-dependent coagulation factor IX that circulates in the blood as an inactive zymogen. This factor is converted to an active form by factor 10 XIa, which excises the activation peptide and thus generates a heavy chain and a light chain held together by one or more disulfide bonds. The role of this activated factor IX in the blood coagulation cascade is to activate factor X to its active form through interactions with Ca⁺² ions, membrane phospholipids, and factor VIII. Alterations of 15 this gene, including point mutations, insertions and deletions, cause factor IX deficiency, which is a recessive X-linked disorder, also called hemophilia B or Christmas disease.

20 **FABP3: fatty acid binding protein 3, muscle and heart (mammary-derived growth inhibitor)**

The intracellular fatty acid-binding proteins (FABPs) belongs to a multigene family. FABPs are divided into at least three distinct types, namely the hepatic-, intestinal- and cardiac-type. They form 14-15 kDa proteins and are thought to participate in the uptake; intracellular metabolism and/or transport of long-chain fatty acids. They may 25 also be responsible in the modulation of cell growth and proliferation. Fatty acid-binding protein 3 gene contains four exons and its function is to arrest growth of mammary epithelial cells. This gene is a candidate tumor suppressor gene for human breast cancer.

FACL3: fatty-acid-Coenzyme A ligase, long-chain 3

The protein encoded by this gene is an isozyme of the long-chain fatty-acid-coenzyme A ligase family. Although differing in substrate specificity, subcellular localization, and tissue distribution, all isozymes of this family convert free long-chain fatty acids into fatty acyl-CoA esters, and thereby play a key role in lipid biosynthesis and fatty acid degradation. This isozyme is highly expressed in brain, and preferentially utilizes myristate, arachidonate, and eicosapentaenoate as substrates. The amino acid sequence of this isozyme is 92% identical to that of rat homolog.

FACL4: fatty-acid-Coenzyme A ligase, long-chain 4

The protein encoded by this gene is an isozyme of the long-chain fatty-acid-coenzyme A ligase family. Although differing in substrate specificity, subcellular localization, and tissue distribution, all isozymes of this family convert free long-chain fatty acids into fatty acyl-CoA esters, and thereby play a key role in lipid biosynthesis and fatty acid degradation. This isozyme preferentially utilizes arachidonate as substrate. The absence of this enzyme may contribute to the mental retardation or Alport syndrome. Alternative splicing of this gene generates 2 transcript variants.

FMO1: flavin containing monooxygenase 1

Metabolic N-oxidation of the diet-derived amino-trimethylamine (TMA) is mediated by flavin-containing monooxygenase and is subject to an inherited FMO3 polymorphism in man resulting in a small subpopulation with reduced TMA N-oxidation capacity resulting in fish odor syndrome Trimethylaminuria. Three forms of the enzyme, FMO1 found in fetal liver, FMO2 found in adult liver, and FMO3 are encoded by genes clustered in the 1q23-q25 region. Flavin-containing

monooxygenases are NADPH-dependent flavoenzymes that catalyzes the oxidation of soft nucleophilic heteroatom centers in drugs, pesticides, and xenobiotics.

GAA: glucosidase, alpha; acid (Pompe disease, glycogen storage disease type II)

5

This gene encodes acid alpha-glucosidase, which is essential for the degradation of glycogen to glucose in lysosomes. Different forms of acid alpha-glucosidase are obtained by proteolytic processing. Defects in this gene are the cause of glycogen storage disease II, also known as Pompe's disease, which is an autosomal recessive disorder with a broad clinical spectrum.

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GAPD: glyceraldehyde-3-phosphate dehydrogenase

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Glyceraldehyde-3-phosphate dehydrogenase catalyzes an important energy-yielding step in carbohydrate metabolism, the reversible oxidative phosphorylation of glyceraldehyde-3-phosphate in the presence of inorganic phosphate and nicotinamide adenine dinucleotide (NAD). The enzyme exists as a tetramer of identical chains. A GAPD pseudogene has been mapped to Xp21-p11 and 15 GAPD-like loci have been identified.

20

GARS: glycyl-tRNA synthetase

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Aminoacyl-tRNA synthetases are a class of enzymes that charge tRNAs with their cognate amino acids. Glycyl-tRNA synthetase is an (alpha)2 dimer which belongs to the class II family of tRNA synthetases. It has been shown to be a target of autoantibodies in the human autoimmune diseases, polymyositis or dermatomyositis.

GBE1: glucan (1,4-alpha-), branching enzyme 1 (glycogen branching enzyme, Andersen disease, glycogen storage disease type IV)

This monomeric enzyme functions in glycogen synthesis by catalyzing the formation of alpha 1,6- glucosidic linkages. It is most highly expressed in liver and muscle. Deficiency can result in glycogen storage disease IV (Andersen's disease).

GP6: glycoprotein VI (platelet)

10 Platelet glycoprotein VI; member of the paired Ig-like receptor family.

GPR-55

Member of the G protein-coupled receptor family.

15 **GPRC5C: G protein-coupled receptor, family C, group 5, member C**

The protein encoded by this gene is a member of the type 3 G protein-coupled receptor family. Members of this superfamily are characterized by a signature 7-transmembrane domain motif. The specific function of this protein is unknown; however, this protein may mediate the cellular effects of retinoic acid on the G protein signal transduction cascade. Alternative splicing in the 5' UTR of this gene results in two transcript variants.

25 **3-hydroxy-3-methylglutaryl coenzyme A synthase**

3-hydroxy-3-methylglutaryl-Coenzyme A synthase; functions in the first step in ketogenesis.

HK1: hexokinase 1

Hexokinases phosphorylate glucose to produce glucose-6-phosphate, thus committing glucose to the glycolytic pathway. This gene encodes a ubiquitous form of hexokinase which localizes to the outer membrane of mitochondria. Mutations in this gene have been associated with hemolytic anemia due to hexokinase deficiency. Alternative splicing of this gene results in five transcript variants which encode different isoforms, some of which are tissue-specific. Each isoform has a distinct N-terminus; the remainder of the protein is identical among all the isoforms. A sixth transcript variant has been described, but due to the presence of several stop codons, it is not thought to encode a protein.

HLA-B associated transcript 3 (BAT3)

A cluster of genes, BAT1-BAT5, has been localized in the vicinity of the genes for TNF alpha and TNF beta. These genes are all within the human major histocompatibility complex class III region. The protein encoded by this gene is a nuclear protein. It has been implicated in the control of apoptosis and regulating heat shock protein. There are three alternatively spliced transcript variants described for this gene.

HMGCL: 3-hydroxymethyl-3-methylglutaryl-Coenzyme A lyase (hydroxymethylglutaricaciduria)

3-Hydroxy-3-methylglutaryl coenzyme A lyase; cleaves 3-OH-3-methylglutaryl CoA to acetoacetic acid and acetyl CoA.

HNF4A: hepatocyte nuclear factor 4, alpha

Nuclear hormone receptor transcription factor; regulates liver specific gene expression.

Chromosome 12 BAC RP11-13J12

Cathepsin B

5

Cathepsin B; lysosomal cysteine (thiol) protease that cleaves APP.

Chromosome 5 clone CTD-2235C13

10 **Chromosome 7 clone RP11-351B12**

Cytochrome P450 3A locus

15 The CYP3A locus includes all the known members of the 3A subfamily of the cytochrome P450 superfamily of genes. These genes encode monooxygenases which catalyze many reactions involved in drug metabolism and synthesis of cholesterol, steroids and other lipids. The CYP3A cluster consists of four genes, CYP3A43, CYP3A4, CYP3A7 and CYP3A5. The region also contains two pseudogenes, CYP3A5P1 and CYP3A5P2, as well as several extra exons which may or may not be included in transcripts produced from this region. Previously another CYP3A member, CYP3A3, was thought to exist; however, it is now thought that this sequence represents a transcript variant of CYP3A4.

20 **ITGB3**

25 The ITGB3 protein product is the integrin beta chain beta 3. Integrins are integral cell-surface proteins composed of an alpha chain and a beta chain. A given chain may combine with multiple partners resulting in different integrins. Integrin beta 3 is found along with the alpha IIb chain in platelets. Integrins are known to participate in cell adhesion as well as cell-surface mediated signalling.

Methionine adenosyltransferase alpha subunit gene fragment.

5 MAT1A encodes methionine adenosyltransferase I (alpha isoform). MAT1A catalyzes the formation of S-adenosylmethionine from methionine and ATP. Both the beta and alpha isoforms may be encoded by MAT1A. Methionine adenosyltransferase deficiency is known to be caused by recessive as well as dominant mutations, the latter identified in autosomal dominant persistent hyper-methioninemia.

10

Homo sapiens PAC clone RP1-102K2 from 22q12.1-qter

Homo sapiens partial ZNF202 gene for zinc finger protein homolog, exon 4

15 Zinc-finger protein 202 may repress genes involved in lipid metabolism; contains zinc fingers.

Homo sapiens vHNF1-C mRNA

Hepatocyte Nuclear Factor 1.

20

Human 2.5 kb mRNA for cytoskeletal tropomyosin TM30(nm)

Human c-kit gene.

25 KIT encodes the human homolog of the proto-oncogene c-kit. 'C-kit' was first identified as the cellular homolog of the feline sarcoma viral oncogene v-kit. KIT is a type 3 transmembrane receptor for MGF (mast cell growth factor, also known as stem cell factor). Mutations in KIT are associated with gastrointestinal stromal tumors, mast cell disease, acute myelogenous leukemia, and piebaldism.

30

Human coagulation factor VII (F7) gene exon 1 and factor X (F10) gene, ex n 1.

This gene encodes coagulation factor VII which is a vitamin K-dependent factor essential for hemostasis. This factor circulates in the blood in a zymogen form, and is converted to an active form by either factor IXa, factor Xa, factor XIIa, or thrombin by minor proteolysis. Upon activation of the factor VII, a heavy chain containing a catalytic domain and a light chain containing 2 EGF-like domains are generated, and two chains are held together by a disulfide bond. In the presence of factor III and calcium ions, the activated factor then further activates the coagulation cascade by converting factor IX to factor IXa and/or factor X to factor Xa. Alternative splicing of this gene results in 2 transcripts. Defects in this gene can cause coagulopathy.

Human cytochrome P450 (CYP1A2) gene, exons 1 and 2.

This gene, CYP1A2, encodes a member of the cytochrome P450 superfamily of enzymes. The cytochrome P450 proteins are monooxygenases which catalyze many reactions involved in drug metabolism and synthesis of cholesterol, steroids and other lipids. The protein encoded by this gene localizes to the endoplasmic reticulum and its expression is induced by some polycyclic aromatic hydrocarbons (PAHs), some of which are found in cigarette smoke. The enzyme's endogenous substrate is unknown; however, it is able to metabolize some PAHs to carcinogenic intermediates. Other xenobiotic substrates for this enzyme include caffeine, aflatoxin B1, and acetaminophen. The transcript from this gene contains four Alu sequences flanked by direct repeats in the 3' untranslated region. A related family member, CYP1A1, is located approximately 25 kb away from CYP1A2 on chromosome .

Human multidrug resistance-associated protein mRNA

See ABCC1.

Human succinyl CoA:3-oxoacid CoA transferase precursor (OXCT) mRNA

The mitochondrial matrix enzyme 3-oxoacid CoA transferase is homodimeric. It is a key enzyme in the extrahepatic utilization of ketone bodies, catalyzing the reversible transfer of coenzyme A from succinyl-CoA to acetoacetate, a necessary step in 5 ketolytic energy production. Deficiencies can result in intermittent ketoacidosis.

Human T-lymphoma invasion and metastasis inducing TIAM1 protein (TIAM1) mRNA

10

Member of the GDP-GTP exchange factor family of proteins; modulates the activity of Rho-like proteins; has a Dbl homology and pleckstrin homology domains.

IL10: interleukin 10

15

Interleukin 10 (cytokine synthesis inhibitory factor); functions as a specific chemotactic factor for CD8+T cells.

IL17R: interleukin 17 receptor

20

Highly similar to murine Il17r; may play a role in T cell activation and induction of IL-2 (Il2).

IL3: interleukin 3 (colony-stimulating factor, multiple)

25

Interleukin-3 (colony-stimulating factor); plays a role in hematopoiesis; member of a family of growth factors.

IL6: interleukin 6 (interferon, beta 2)

Interleukin 6 (interferon-beta 2); induces the maturation of B cells into immunoglobulin-secreting cells.

5

IL8RA: interleukin 8 receptor, alpha

Interleukin 8 receptor alpha; G protein-coupled receptor that mediates neutrophil chemotaxis and binds interleukin 8 (IL8).

10

INHBC: inhibin, beta C

This gene encodes the beta C chain of inhibin, a member of the TGF-beta superfamily. This subunit forms heterodimers with beta A and beta B subunits.

15

Inhibins and activins, also members of the TGF-beta superfamily, are hormones with opposing actions and are involved in hypothalamic, pituitary, and gonadal hormone secretion, as well as growth and differentiation of various cell types.

20

ITGAL: integrin, alpha L (antigen CD11A (p180), lymphocyte function-associated antigen 1; alpha polypeptide)

25

ITGAL encodes the integrin alpha L chain. Integrins are heterodimeric integral membrane proteins composed of an alpha chain and a beta chain. This I-domain containing alpha integrin combines with the beta 2 chain (ITGB2) to form the integrin lymphocyte function-associated antigen-1 (LFA-1), which is expressed on all leukocytes. LFA-1 plays a central role in leukocyte intercellular adhesion through interactions with its ligands, ICAMs 1-3 (intercellular adhesion molecules 1 through 3), and also functions in lymphocyte costimulatory signaling.

ITGB2: integrin, beta 2 (antigen CD18 (p95), lymphocyte function-associated antigen 1; macrophage antigen 1 (mac-1) beta subunit)

5 The ITGB2 protein product is the integrin beta chain beta 2. Integrins are integral cell-surface proteins composed of an alpha chain and a beta chain. A given chain may combine with multiple partners resulting in different integrins. For example, beta 2 combines with the alpha L chain to form the integrin LFA-1, and combines with the alpha M chain to form the integrin Mac-1. Integrins are known to participate in cell adhesion as well as cell-surface mediated signalling.

10

KCNQ1: potassium voltage-gated channel, KQT-like subfamily, member 1

15 KCNQ1 encodes the K⁺ channel subunit responsible for the delayed-rectifier K⁺ current in cardiac myocytes. The delayed-rectifier channel is completed by the protein encoded by KCNE1. Mutations in KCNQ1 cause inherited long-QT syndrome.

LAMA3: laminin, alpha 3 (nicaerin (150kD), kalinin (165kD), BM600 (150kD), epilegrin)

20

25 Laminins are basement membrane components thought to mediate the attachment, migration and organization of cells into tissues during embryonic development by interacting with other extracellular matrix components. The protein encoded by this gene is the alpha-3 chain of laminin 5, which is a complex glycoprotein composed of three subunits (alpha, beta, and gamma). Laminin 5 is thought to be involved in cell adhesion, signal transduction and differentiation of keratinocytes. Mutations in this gene have been identified as the cause of Herlitz type junctional epidermolysis bullosa. Alternative splicing has been observed at this locus but the full-length nature of these variants has not been determined.

30

LAMR1: laminin receptor 1 (67kD, ribosomal protein SA)

Laminins, a family of extracellular matrix glycoproteins, are the major non-collagenous constituent of basement membranes. They have been implicated in a wide variety of biological processes including cell adhesion, differentiation, migration, signaling, neurite outgrowth and metastasis. Many of the effects of laminin are mediated through interactions with cell surface receptors. These receptors include members of the integrin family, as well as non-integrin laminin-binding proteins. This gene encodes a high-affinity, non-integrin family, laminin receptor 1.

This receptor has been variously called 67 kD laminin receptor, 37 kD laminin receptor precursor (37LRP) and p40 ribosome-associated protein. The amino acid sequence of laminin receptor 1 is highly conserved through evolution, suggesting a key biological function. It has been observed that the level of the laminin receptor transcript is higher in colon carcinoma tissue and lung cancer cell line than their normal counterparts. Also, there is a correlation between the upregulation of this polypeptide in cancer cells and their invasive and metastatic phenotype. Multiple copies of this gene exist, however, most of them are pseudogenes thought to have arisen from retropositional events..

LDLR: low density lipoprotein receptor (familial hypercholesterolemia)

The low density lipoprotein receptor (LDLR) gene family consists of cell surface proteins involved in receptor-mediated endocytosis of specific ligands. Low density lipoprotein (LDL) is normally bound at the cell membrane and taken into the cell ending up in lysosomes where the protein is degraded and the cholesterol is made available for repression of microsomal enzyme 3-hydroxy-3-methylglutaryl coenzyme A (HMG CoA) reductase, the rate-limiting step in cholesterol synthesis. At the same time, a reciprocal stimulation of cholesterol ester synthesis takes place. Mutations in the LDL receptor (LDLR) gene cause the autosomal dominant disorder, familial hypercholesterolemia.

LGALS7: lectin, galactoside-binding, soluble, 7 (galectin 7)

The galectins are a family of beta-galactoside-binding proteins implicated in modulating cell-cell and cell-matrix interactions. Differential and *in situ* hybridizations indicate that this lectin is specifically expressed in keratinocytes. It is expressed at all stages of epidermal differentiation (i.e., in basal and suprabasal layers). It is moderately repressed by retinoic acid. The protein was found mainly in stratified squamous epithelium. The antigen localized to basal keratinocytes, although it was also found, albeit at lower levels, in the suprabasal layers where it concentrated to areas of cell-to-cell contact. The cellular localization and its striking down-regulation in cultured keratinocytes imply a role in cell-cell and/or cell-matrix interactions necessary for normal growth control.

LIMK1: LIM domain kinase 1

15

There are approximately 40 known eukaryotic LIM proteins, so named for the LIM domains they contain. LIM domains are highly conserved cysteine-rich structures containing 2 zinc fingers. Although zinc fingers usually function by binding to DNA or RNA, the LIM motif probably mediates protein-protein interactions. LIM kinase-1 and LIM kinase-2 belong to a small subfamily with a unique combination of 2 N-terminal LIM motifs and a C-terminal protein kinase domain. LIMK1 is likely to be a component of an intracellular signaling pathway and may be involved in brain development. LIMK1 hemizygosity is implicated in the impaired visuospatial constructive cognition of Williams syndrome. Two splice variant have been identified.

LMNB2: lamin B2

Lamin B2; member of a family of structural nuclear envelope proteins.

30

LPL: lipoprotein lipase

LPL encodes lipoprotein lipase, which is expressed in heart, muscle, and adipose tissue. LPL functions as a homodimer, and has the dual functions of triglyceride hydrolase and ligand/bridging factor for receptor-mediated lipoprotein uptake. Severe mutations that cause LPL deficiency result in type I hyperlipoproteinemia, while less extreme mutations in LPL are linked to many disorders of lipoprotein metabolism.

LRP8: low density lipoprotein receptor-related protein 8, apolipoprotein e receptor

This gene encodes an apolipoprotein E receptor, a member of the low density lipoprotein receptor (LDLR) family. Apolipoprotein E is a small lipophilic plasma protein and a component of lipoproteins such as chylomicron remnants, very low density lipoprotein (VLDL), and high density lipoprotein (HDL). The apolipoprotein E receptor is involved in cellular recognition and internalization of these lipoproteins. Alternative splicing generates three transcript variants for this gene; additional variants have been described, but their full length nature has not been determined.

LSS: lanosterol synthase (2,3-oxidosqualene-lanosterol cyclase)

Lanosterol synthase ((S)-2,3-epoxysqualene mutase); catalyzes the cyclization of (S)-2,3-oxidosqualene; forms lanosterol during sterol biosynthesis.

LTA: lymphotoxin alpha (TNF superfamily, member 1)

Lymphotoxin alpha, a member of the tumor necrosis factor family, is a cytokine produced by lymphocytes. LTA is highly inducible, secreted, and exists as homotrimeric molecule. LTA forms heterotrimers with lymphotoxin-beta which anchors lymphotoxin-alpha to the cell surface. LTA mediates a large variety of inflammatory, immunostimulatory, and antiviral responses. LTA is also involved in

the formation of secondary lymphoid organs during development and plays a role in apoptosis.

MAOA: monoamine oxidase A

5

MAOA encodes monoamine oxidase A, an enzyme that degrades amine neurotransmitters, such as dopamine, norepinephrine, and serotonin. Deficiency of this enzyme results in Brunner syndrome.

10

MARCKS: myristoylated alanine-rich protein kinase C substrate

The protein encoded by this gene is a substrate for protein kinase C. It is localized to the plasma membrane and is an actin filament crosslinking protein. Phosphorylation by protein kinase C or binding to calcium-calmodulin inhibits its association with actin and with the plasma membrane, leading to its presence in the cytoplasm. The protein is thought to be involved in cell motility, phagocytosis, membrane trafficking and mitogenesis.

MCL1: myeloid cell leukemia sequence 1 (BCL2-related)

20

Similar to BCL2.

MCP: membrane cofactor protein (CD46, trophoblast-lymphocyte cross-reactive antigen)

25

Membrane cofactor protein; acts as the receptor for the measles virus, may be involved in the regulation of complement activation; contains SCRs.

METTL1: methyltransferase-like 1

This gene is an ortholog of the *S. cerevisiae* YDL201w gene, which is predicted to encode a methyltransferase. The gene product contains a conserved S-adenosyl-methionine-binding motif, which is typical of a methyltransferase. Alternative splice variants encoding different protein isoforms and transcript variants utilizing alternative polyA sites have been described in the literature.

5 **MLLT3: myeloid/lymphoid or mixed-lineage leukemia (trithorax homolog,
10 Drosophila);**

Serine and proline rich protein, has a nuclear targeting sequence.

15 **MTHFD1: methylenetetrahydrofolate dehydrogenase (NADP+ dependent),
methenyltetrahydrofolate cyclohydrolase, formyltetrahydrofolate synthetase**

20 This gene encodes a protein that possesses three distinct enzymatic activities, 5,10-methylenetetrahydrofolate dehydrogenase, 5,10-methenyltetrahydrofolate cyclohydrolase and 10-formyltetrahydrofolate synthetase. Each of these activities catalyzes one of three sequential reactions in the interconversion of 1-carbon derivatives of tetrahydrofolate, which are substrates for methionine, thymidylate, and de novo purine syntheses. The trifunctional enzymatic activities are conferred by two major domains, an aminoterminal portion containing the dehydrogenase and cyclohydrolase activities and a larger synthetase domain.

MTMR2 myotubularin related protein 2 (MTMR2)

This gene is a member of the myotubularin family and encodes a putative tyrosine phosphatase. Mutations in this gene are a cause of Charcot-Marie-Tooth disease type 5 4B, an autosomal recessive demyelinating neuropathy. This gene utilizes multiple polyA signals, only one of which has been determined.

Muscle specific serine kinase (MSSK1; serine/threonine kinase 23, STK23),

10 Highly similar to SRPK2; may be protein kinase for SR family of RNA splicing factors; contains a kinase domain.

MVD: mevalonate (diphospho) decarboxylase

15 The enzyme mevalonate pyrophosphate decarboxylase catalyzes the conversion of mevalonate pyrophosphate into isopentenyl pyrophosphate in one of the early steps in cholesterol biosynthesis. It decarboxylates and dehydrates its substrate while hydrolyzing ATP.

20 **MYH11: myosin, heavy polypeptide 11, smooth muscle**

The protein encoded by this gene is a smooth muscle myosin belonging to the myosin heavy chain family. The gene product is a subunit of a hexameric protein that consists of 2 heavy chain subunits and 2 pairs of non-identical light chain subunits. It 25 functions as a major contractile protein, converting chemical energy into mechanical energy through the hydrolysis of ATP. The gene encoding a human ortholog of rat NUDE1 is transcribed from the reverse strand of MYH11 gene, and its 3' end overlaps with that of the latter. The pericentric inversion of chromosome 16 [inv(16)(p13q22)] produces a chimeric transcript consisting of the first 165 residues 30 from the N terminus of core-binding factor beta in a fusion with the C-terminal portion of the smooth muscle myosin heavy chain. This chromosomal rearrangement

is associated with acute myeloid leukemia of the M4Eo subtype. Alternative splicing generates isoforms that are differentially expressed, with ratios changing during muscle cell maturation. Additional splice variants have been described but their full-length nature has not been determined..

5

MYH7: myosin, heavy polypeptide 7, cardiac muscle, beta

MYH7 encodes the cardiac muscle beta (or slow) isoform of myosin. Changes in the relative abundance of MYH7 and MYH6 (the alpha, or fast, isoform of cardiac myosin heavy chain) correlate with the contractile velocity of cardiac muscle. Mutations in MYH7 are associated with familial hypertrophic cardiomyopathy.

NADH dehydrogenase (ubiquinone) 1, alpha subcomplex, 4 (9kD, MLRQ), NDUFA4

15

Subunit of NADH-ubiquinone oxidoreductase (complex I); transports electrons from NADH to ubiquinone.

NADH-UBIQUINONE OXIDOREDUCTASE CHAIN 5 (EC 1:6.5.3).

20

Subunit of NADH-ubiquinone oxidoreductase (complex I); transports electrons from NADH to ubiquinone.

NDUFA9: NADH dehydrogenase (ubiquinone) 1 alpha subcomplex, 9 (39kD)

25

NGFB: nerve growth factor, beta polypeptide

Nerve growth factor beta; has roles in neuronal differentiation and survival.

NGFR: nerve growth factor receptor (TNFR superfamily, member 16)

Nerve growth factor receptor contains an extracellular domain containing four 40-amino acid repeats with 6 cysteine residues at conserved positions followed by a serine/threonine-rich region, a single transmembrane domain, and a 155-amino acid cytoplasmic domain. The cysteine-rich region contains the nerve growth factor binding domain.

NID2: nidogen 2

10

Nidogen-2; basement membrane protein.

HSU15552: acidic 82 kDa protein mRNA

15

Nonmuscle type myosin heavy chain 9 (MYH9)

Non-muscle myosin heavy chain 9; motor protein that provides force for muscle contraction, cytokinesis and phagocytosis; contains an ATPase head domain and a rod-like tail domain.

20

NPC1: Niemann-Pick disease, type C1

25

NPC1 was identified as the gene that when mutated, results in Niemann-Pick C disease. NPC1 encodes a putative integral membrane protein containing motifs consistent with a role in intracellular transport of cholesterol to post-lysosomal destinations.

Nth endonuclease III-like 1 (NTHL1)

30

Endonuclease; excises damaged pyrimidines.

NUCB2: nucleobindin 2

Nucleobindin 2; may bind DNA and calcium; has DNA-binding and EF-hand domains, and a leucine-zipper.

5

nuclear receptor subfamily 1, group I, member 2 (NR1I2)

The gene product belongs to the nuclear receptor superfamily, members of which are transcription factors characterized by a ligand-binding domain and a DNA-binding domain. The encoded protein is a transcriptional regulator of the cytochrome P450 gene CYP3A4, binding to the response element of the CYP3A4 promoter as a heterodimer with the 9-cis retinoic acid receptor RXR. It is activated by a range of compounds that induce CYP3A4, including dexamethasone and rifampicin. The gene product contains a zinc finger domain. Three alternatively spliced transcripts that encode different isoforms have been described, one of which encodes two products through the use of alternative translation initiation codons. Additional transcript variants derived from alternative promoter usage, alternative splicing, and/or alternative polyadenylation exist, but they have not been fully described.

20

OGDH: oxoglutarate (alpha-ketoglutarate) dehydrogenase (lipoamide)

Alpha-ketoglutarate or 2-oxoglutarate dehydrogenase; helps convert a-ketoglutarate to succinyl coenzyme A in Krebs cycle.

25

OXCT: 3-oxoacid CoA transferase

30

The mitochondrial matrix enzyme 3-oxoacid CoA transferase is homodimeric. It is a key enzyme in the extrahepatic utilization of ketone bodies, catalyzing the reversible transfer of coenzyme A from succinyl-CoA to acetoacetate, a necessary step in ketolytic energy production. Deficiencies can result in intermittent ketoacidosis.

P2RY1: purinergic receptor P2Y, G-protein coupled, 1

Purinergic receptor P2Y1, a G protein-coupled receptor; mediates responses to ATP and increases inositol phosphate levels.

5

PCCA: propionyl Coenzyme A carboxylase, alpha polypeptide

PCCA encodes the alpha subunit of the heterodimeric mitochondrial enzyme Propionyl-CoA carboxylase. PCCA encodes the biotin-binding region of this enzyme. Mutations in either PCCA or PCCB (encoding the beta subunit) lead to an enzyme deficiency result in propionic acidemia.

10

PDGFB: platelet-derived growth factor beta polypeptide (simian sarcoma viral (v-sis) oncogene homolog)

15

The protein encoded by this gene is a member of the platelet-derived growth factor family. The four members of this family are mitogenic factors for cells of mesenchymal origin and are characterized by a motif of eight cysteines. This gene product can exist either as a homodimer or as a heterodimer with the platelet-derived growth factor alpha polypeptide, where the dimers are connected by disulfide bonds. Mutations in this gene are associated with meningioma. Reciprocal translocations between chromosomes 22 and 7, at sites where this gene and that for COL1A1 are located, are associated with a particular type of skin tumor called dermatofibrosarcoma protuberans resulting from unregulated expression of growth factor. Two splice variants have been identified for this gene.

20

25

PERIOD CIRCADIAN PROTEIN 2 (KIAA0347)

30

This gene is a member of the Period family of genes and is expressed in a circadian pattern in the suprachiasmatic nucleus, the primary circadian pacemaker in the mammalian brain. Genes in this family encode components of the circadian rhythms

of locomotor activity, metabolism, and behavior. Circadian expression in the suprachiasmatic nucleus continues in constant darkness, and a shift in the light/dark cycle evokes a proportional shift of gene expression in the suprachiasmatic nucleus. The specific function of this gene is not yet known.

5

Peroxisome proliferative activated receptor, delta (PPARD)

Peroxisome proliferator-activated receptor delta is a member of the steroid hormone receptor superfamily.

10

PGM5: phosphoglucomutase 5

Phosphoglucomutase-related (aciculin) putative structural protein; interacts with the cytoskeletal proteins dystrophin and utrophin.

15

PLA2G3: phospholipase A2, group III

Group III secreted phospholipase A2; calcium-dependent, displays a preference for phosphatidylglycerol over phosphatidylcholine.

20

PLA2G4C: phospholipase A2, group IVC (cytosolic, calcium-independent)

Group IVC calcium-independent phospholipase a2; hydrolyzes the phospholipid sn-2 ester bond; member of the phospholipase family.

25

PLA2G6: phospholipase A2, group VI (cytosolic, calcium-independent)

Cytosolic calcium-independent phospholipase_a2; hydrolyzes the phospholipid sn-2 ester bond; member of the phospholipase family.

30

PMVK: phosphomevalonate kinase

Phosphomevalonate kinase; converts mevalonate-5-phosphate to mevalonate-5-diphosphate.

5

PNMT: phenylethanolamine N-methyltransferase

Phenylethanolamine N-methyltransferase; converts norepinephrine to epinephrine.

10 **PON1: paraoxonase 1**

PON2: paraoxonase 2

Paraoxonase/arylesterase 2; possibly functions in protecting low density lipoprotein against oxidative modification; member of a family that hydrolyzes toxic organophosphates.

PPARA: peroxisome proliferative activated receptor, alpha

20 Peroxisome proliferators are a diverse group of chemicals which include hypolipidemic drugs, herbicides, leukotriene antagonists, and plasticizers, and are so called because they induce an increase in the size and number of peroxisomes. Peroxisomes are subcellular organelles found in plants and animals, and contain enzymes for respiration, cholesterol and lipid metabolism. Infact, the fibrate class of
25 hypolipidemic drugs is used to reduce triglycerides and cholesterol in patients with hyperlipidemia, a major risk factor for coronary heart disease. The action of peroxisome proliferators is thought to be mediated via specific receptors belonging to the steroid hormone receptor superfamily, called PPARs. Thus far, four closely related subtypes, alpha, beta, gamma and delta, have been identified. The subtype
30 PPAR-alpha, encoded by PPARA, is a nuclear transcription factor. Upon activation

by peroxisome proliferators, it modulates the expression of target genes involved in lipid metabolism, suggesting a role for PPAR-alpha in lipid homeostasis.

PPARG: peroxisome proliferative activated receptor, gamma

5

The protein encoded by this gene is a member of the peroxisome proliferator-activated receptor (PPAR) subfamily of nuclear receptors. PPARs form heterodimers with retinoid X receptors (RXRs) and these heterodimers regulate transcription of various genes. Three subtypes of PPARs are known: PPAR-alpha, PPAR-delta, and PPAR-gamma. The protein encoded by this gene is PPAR-gamma and is a regulator of adipocyte differentiation. Additionally, PPAR-gamma has been implicated in the pathology of numerous diseases including obesity, diabetes, atherosclerosis and cancer. Multiple transcript variants that use alternate promoters and splicing have been identified for this gene. At least three of these variants encode the same isoform.

10

15

PPM1A: protein phosphatase 1A (formerly 2C), magnesium-dependent, alpha isoform

20

Magnesium- or manganese-dependent alpha protein phosphatase 1A; regulates cell stress responses.

PROBABLE G PROTEIN-COUPLED RECEPTOR APJ.

25

PTPRA: protein tyrosine phosphatase, receptor type, A

30

The protein encoded by this gene is a member of the protein tyrosine phosphatase (PTP) family. PTPs are known to be signaling molecules that regulate a variety of cellular processes including cell growth, differentiation, mitotic cycle, and oncogenic transformation. This PTP contains an extracellular domain, a single transmembrane segment and two tandem intracytoplasmic catalytic domains, and thus represents a

receptor-type PTP. This PTP has been shown to dephosphorylate and activate Src family tyrosine kinases, and is implicated in the regulation of integrin signaling, cell adhesion and proliferation. Three alternatively spliced variants of this gene, which encode two distinct isoforms, have been reported.

5

PYGM: phosphorylase, glycogen; muscle (McArdle syndrome, glycogen storage disease type V)

Muscle glycogen phosphorylase

10

RTN1: reticulon 1

RXRA: retinoid X receptor, alpha

15 Retinoid X receptors (RXRs) and retinoic acid receptors (RARs), are nuclear receptors that mediate the biological effects of retinoids by their involvement in retinoic acid-mediated gene activation. These receptors exert their action by binding, as homodimers or heterodimers, to specific sequences in the promoters of target genes and regulating their transcription. The protein encoded by this gene is a
20 member of the steroid and thyroid hormone receptor superfamily of transcriptional regulators.

RXRB: retinoid X receptor, beta

25 Retinoid X receptor beta; binds to and serves as transcriptional coactivator for retinoic acid.

SCA1: spinocerebellar ataxia 1 (olivopontocerebellar ataxia 1, autosomal dominant, ataxin 1)

The autosomal dominant cerebellar ataxias (ADCA) are a heterogeneous group of neurodegenerative disorders characterized by progressive degeneration of the cerebellum, brain stem and spinal cord. Clinically, ADCA has been divided into three groups: ADCA types I-III. ADCAI is genetically heterogeneous, with five genetic loci, designated spinocerebellar ataxia (SCA) 1, 2, 3, 4 and 6, being assigned to five different chromosomes. ADCAI, which always presents with retinal degeneration (SCA7), and ADCAIII often referred to as the 'pure' cerebellar syndrome (SCA5), are most likely homogeneous disorders. Several SCA genes have been cloned and shown to contain CAG repeats in their coding regions. ADCA is caused by the expansion of the CAG repeats, producing an elongated polyglutamine tract in the corresponding protein. The expanded repeats are variable in size and unstable, usually increasing in size when transmitted to successive generations. The function of the ataxins is not known. The SCA1 locus has been mapped to chromosome 6, and it has been determined that the diseased allele contains 41-81 CAG repeats, compared to 6-39 in the normal allele. Several transcript variants of SCA1 in the 5' UTR have been described; however, their full-length nature is not known..

SDF1: stromal cell-derived factor 1

Stromal cell-derived factor 1; lymphocyte chemoattractant that signals through the receptor CXCR4.

SERPINA5: serine (or cysteine) proteinase inhibitor, clade A (alpha-1 antiproteinase, antitrypsin), member 5

Protein C inhibitor (plasminogen activator inhibitor III); may be a serine protease inhibitor; member of the serpin family of serine protease inhibitors.

SERPINH1: serine (or cysteine) proteinase inhibitor, clade H (heat shock protein 47), member 1, (collagen binding protein 1)

5 Colligin; collagen-binding protein; Similar to HSPs and to serpin family serine protease inhibitors.

SLC21A6: solute carrier family 21 (organic anion transporter), member 6

10 Organic anion transporter.

SLC27A1: solute carrier family 27 (fatty acid transporter), member 1

SULT1A2: sulfotransferase family, cytosolic, 1A, phenol-preferring, member 2

15 Phenol-metabolizing sulfotransferase 2; sulfonates simple planar phenols.

THBS3: Thrombospondin 3

Thrombospondin 3 binds heparin and calcium; similar to murine Thbs3

20 **TBP: TATA box binding protein**

TATA box binding protein, component of the TFIID complex; functions in the initiation of mRNA synthesis and basal transcription.

25 **TBXA2R: thromboxane A2 receptor**

Thromboxane A2 receptor (prostaglandin H2 receptor); G protein-coupled receptor, activates Ca²⁺-activated chloride channels; stimulates platelet aggregation and smooth muscle constriction.

TCF2: transcription factor 2, hepatic; LF-B3; variant hepatic nuclear factor

TCF2 encodes transcription factor 2, a liver-specific factor of the homeobox-containing basic helix-turn-helix family. The TCF2 protein is believed to form heterodimers with another liver-specific member of this transcription factor family, TCF1; depending on the TCF2 isoform, the result may be to activate or inhibit transcription of target genes. Mutation of TCF2 that disrupts normal function has been identified as the cause of MODY5 (Maturity-Onset of Diabetes, Type 5). A third human transcript variant is believed to exist based on such a variant in the rat; however, to date such an mRNA species has not been isolated.

TETRAN: tetracycline transporter-like protein

Similar to E. coli tetracycline resistance efflux protein.

15

TGFB1: transforming growth factor, beta 1 (Camurati-Engelmann disease)

Transforming growth factor-beta 1; regulates cell proliferation, differentiation, and apoptosis.

20

TGFB2: transforming growth factor, beta 2

Transforming growth factor-beta 2 (glioblastoma-derived T cell suppressor factor); suppresses IL2 - dependent growth of T cells; member of a family of cytokines that transmits signals through transmembrane serine/threonine kinases.

25

TGFB3: transforming growth factor, beta 3

Transforming growth factor-beta 3; transmits signals through transmembrane serine/threonine kinases, may be required for normal development of the lung and palate; member of family of cytokines, very strongly similar to murine Tgfb3.

30

THPO: thrombopoietin (myeloproliferative leukemia virus oncogene ligand, megakaryocyte growth and development factor)

- 5 Thrombopoietin; binds to c-Mpl receptor and regulates megakaryocyte development.

TNFAIP2: tumor necrosis factor, alpha-induced protein 2

- Secreted by vascular endothelium, expression is induced by tumor necrosis factor
10 alpha, interleukin-1 beta, and lipopolysaccharide.

TRAP1: heat shock protein 75

- Heat shock protein 75; binds and refolds denatured RB1 during M phase and after
15 heat shock; member of the HSP90 family of molecular chaperones.

TRIP10: thyroid hormone receptor interactor 10

- Similar to the non-kinase domains of FER and Fes/Fps tyrosine kinases; binds to
20 activated Cdc42 and may regulate actin cytoskeleton; contains an SH3 domain.

TXN: thioredoxin

Thioredoxin; has dithiol-disulfide oxidoreductase activity.

25

USP6: ubiquitin specific protease 6 (Tre-2 oncogene)

Ubiquitin specific protease 6 (Tre-2 oncogene); cleaves ubiquitin from proteins, has predicted nucleic acid-binding properties.

30

UTRN: utr phin (homologous to dystrophin)

This gene shares both structural and functional similarities with the dystrophin gene. It contains an actin-binding N-terminus, a triple coiled-coil repeat central region, and a C-terminus that consists of protein-protein interaction motifs which interact with dystroglycan protein components. The protein encoded by this gene is located at the neuromuscular synapse and myotendinous junctions, where it participates in post-synaptic membrane maintenance and acetylcholine receptor clustering. Mouse studies suggest that this gene may serve as a functional substitute for the dystrophin gene and therefore, may serve as a potential therapeutic alternative to muscular dystrophy which caused by mutations in the dystrophin gene. Alternative splicing of the utrophin gene has been described; however, the full-length nature of these variants has not yet been determined.

15 VEGF: vascular endothelial growth factor

Vascular endothelial growth factor; induces endothelial cell proliferation and vascular permeability.

20 VEGFB: vascular endothelial growth factor B

Vascular endothelial growth factor B; involved in angiogenesis and endothelial cell growth.

25 WISP1: WNT1 inducible signaling pathway protein 1

This gene encodes a member of the WNT1 inducible signaling pathway (WISP) protein subfamily, which belongs to the connective tissue growth factor (CTGF) family. WNT1 is a member of a family of cysteine-rich, glycosylated signaling proteins that mediate diverse developmental processes. The CTGF family members are characterized by four conserved cysteine-rich domains: insulin-like growth

factor-binding domain, von Willebrand factor type C module, thrombospondin domain and C-terminal cystine knot-like domain. This gene may be downstream in the WNT1 signaling pathway that is relevant to malignant transformation. It is expressed at a high level in fibroblast cells, and overexpressed in colon tumors. The
5 encoded protein binds to decorin and biglycan, two members of a family of small leucine-rich proteoglycans present in the extracellular matrix of connective tissue, and possibly prevents the inhibitory activity of decorin and biglycan in tumor cell proliferation. It also attenuates p53-mediated apoptosis in response to DNA damage through activation of the Akt kinase. It is 83% identical to the mouse protein at the
10 amino acid level. Alternative splicing of this gene generates 2 transcript variants..

XDH: xanthene dehydrogenase

Xanthine dehydrogenase belongs to the group of molybdenum-containing hydroxylases involved in the oxidative metabolism of purines. The enzyme is a homodimer. Xanthine dehydrogenase can be converted to xanthine oxidase by reversible sulphydryl oxidation or by irreversible proteolytic modification. Defects in xanthine dehydrogenase cause xanthinuria, may contribute to adult respiratory distress syndrome, and may potentiate influenza infection through an oxygen metabolite-dependent mechanism.
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20

YAP1: Yes-associated protein 1, 65 kD

Yes-associated protein; binds to the proto-oncoprotein Yes; has a WW domain.

PROCR: protein C receptor, endothelial (EPCR)

Endothelial Protein C receptor; binds protein C in a calcium-dependent manner; member of the CD1/major histocompatibility complex superfamily.

STX1A: syntaxin 1A (brain)

30 Syntaxin 1A (brain); involved in intracellular transport and neurotransmitter release.

As SNPs are linked to other SNPs in neighboring genes on a chromosome (Linkage Disequilibrium) those SNPs could also be used as marker SNPs. In a recent publication it was shown that SNPs are linked over 100 kb in some cases more than 150 kb (Reich D.E. et al. Nature 411, 199-204, 2001). Hence SNPs lying in regions neighbouring PA SNPs could be linked to the latter and by this being a diagnostic marker. These associations could be performed as described for the gene polymorphism in methods.

Definitions

10

For convenience, the meaning of certain terms and phrases employed in the specification, examples, and appended claims are provided below. Moreover, the definitions by itself are intended to explain a further background of the invention.

15

The term "allele", which is used interchangeably herein with "allelic variant" refers to alternative forms of a gene or portions thereof. Alleles occupy the same locus or position on homologous chromosomes. When a subject has two identical alleles of a gene, the subject is said to be homozygous for the gene or allele. When a subject has two different alleles of a gene, the subject is said to be heterozygous for the gene. 20 Alleles of a specific gene can differ from each other in a single nucleotide, or several nucleotides, and can include substitutions, deletions, and insertions of nucleotides. An allele of a gene can also be a form of a gene containing a mutation.

25

The term "allelic variant of a polymorphic region of a gene" refers to a region of a gene having one of several nucleotide sequences found in that region of the gene in other individuals.

30

"Homology" or "identity" or "similarity" refers to sequence similarity between two peptides or between two nucleic acid molecules. Homology can be determined by comparing a position in each sequence which may be aligned for purposes of comparison. When a position in the compared sequence is occupied by the same base

or amino acid, then the molecules are homologous at that position. A degree of homology between sequences is a function of the number of matching or homologous positions shared by the sequences. An "unrelated" or "non-homologous" sequence shares less than 40% identity, though preferably less than 25% identity,
5 with one of the sequences of the present invention.

The term "a homologue of a nucleic acid" refers to a nucleic acid having a nucleotide sequence having a certain degree of homology with the nucleotide sequence of the nucleic acid or complement thereof. A homologue of a double stranded-nucleic acid
10 having SEQ ID NO. X is intended to include nucleic acids having a nucleotide sequence which has a certain degree of homology with SEQ ID NO. X or with the complement thereof. Preferred homologous of nucleic acids are capable of hybridizing to the nucleic acid or complement thereof.

15 The term "interact" as used herein is meant to include detectable interactions between molecules, such as can be detected using, for example, a hybridization assay.

The term interact is also meant to include "binding" interactions between molecules.
Interactions may be, for example, protein-protein, protein-nucleic acid, protein-small
20 molecule or small molecule-nucleic acid in nature.

The term "intronic sequence" or "intronic nucleotide sequence" refers to the nucleotide sequence of an intron or portion thereof.

25 The term "isolated" as used herein with respect to nucleic acids, such as DNA or RNA, refers to molecules separated from other DNAs or RNAs, respectively, that are present in the natural source of the macromolecule. The term isolated as used herein also refers to a nucleic acid or peptide that is substantially free of cellular material, viral material, or culture medium when produced by recombinant DNA techniques,
30 or chemical precursors or other chemicals when chemically synthesized.

Moreover, an "isolated nucleic acid" is meant to include nucleic acid fragments which are not naturally occurring as fragments and would not be found in the natural state. The term "isolated" is also used herein to refer to polypeptides which are isolated from other cellular proteins and is meant to encompass both purified and recombinant polypeptides.

The term "lipid" shall refer to a fat or fat-like substance that is insoluble in polar solvents such as water. The term "lipid" is intended to include true fats (e.g. esters of fatty acids and glycerol); lipids (phospholipids, cerebrosides, waxes); sterols (cholesterol, ergosterol) and lipoproteins (e.g. HDL, LDL and VLDL).

The term "locus" refers to a specific position in a chromosome. For example, a locus of a gene refers to the chromosomal position of the gene.

The term "modulation" as used herein refers to both up-regulation, (i.e., activation or stimulation), for example by agonizing, and down-regulation (i.e. inhibition or suppression), for example by antagonizing of a bioactivity (e.g. expression of a gene).

The term "molecular structure" of a gene or a portion thereof refers to the structure as defined by the nucleotide content (including deletions, substitutions, additions of one or more nucleotides), the nucleotide sequence, the state of methylation, and/or any other modification of the gene or portion thereof.

The term "mutated gene" refers to an allelic form of a gene, which is capable of altering the phenotype of a subject having the mutated gene relative to a subject which does not have the mutated gene. If a subject must be homozygous for this mutation to have an altered phenotype, the mutation is said to be recessive. If one copy of the mutated gene is sufficient to alter the genotype of the subject, the mutation is said to be dominant. If a subject has one copy of the mutated gene and

has a phenotype that is intermediate between that of a homozygous and that of a heterozygous (for that gene) subject, the mutation is said to be co-dominant.

As used herein, the term "nucleic acid" refers to polynucleotides such as deoxyribonucleic acid (DNA), and, where appropriate, ribonucleic acid (RNA). The term should also be understood to include, as equivalents, derivatives, variants and analogs of either RNA or DNA made from nucleotide analogs, including peptide nucleic acids (PNA), morpholino oligonucleotides (J. Summerton and D. Weller, Antisense and Nucleic Acid Drug Development 7:187 (1997)) and, as applicable to the embodiment being described, single (sense or antisense) and double-stranded polynucleotides. Deoxyribonucleotides include deoxyadenosine, deoxycytidine, deoxyguanosine, and deoxythymidine. For purposes of clarity, when referring herein to a nucleotide of a nucleic acid, which can be DNA or an RNA, the term "adenosine", "cytidine", "guanosine", and "thymidine" are used. It is understood that if the nucleic acid is RNA, a nucleotide having a uracil base is uridine.

The term "nucleotide sequence complementary to the nucleotide sequence set forth in SEQ ID NO. x" refers to the nucleotide sequence of the complementary strand of a nucleic acid strand having SEQ ID NO. x. The term "complementary strand" is used herein interchangeably with the term "complement". The complement of a nucleic acid strand can be the complement of a coding strand or the complement of a non-coding strand. When referring to double stranded nucleic acids, the complement of a nucleic acid having SEQ ID NO. x refers to the complementary strand of the strand having SEQ ID NO. x or to any nucleic acid having the nucleotide sequence of the complementary strand of SEQ ID NO. x. When referring to a single stranded nucleic acid having the nucleotide sequence SEQ ID NO. x, the complement of this nucleic acid is a nucleic acid having a nucleotide sequence which is complementary to that of SEQ ID NO. x. The nucleotide sequences and complementary sequences thereof are always given in the 5' to 3' direction. The term "complement" and "reverse complement" are used interchangeably herein.

The term "operably linked" is intended to mean that the promoter is associated with the nucleic acid in such a manner as to facilitate transcription of the nucleic acid.

5 The term "polymorphism" refers to the coexistence of more than one form of a gene or portion thereof. A portion of a gene of which there are at least two different forms, i.e., two different nucleotide sequences, is referred to as a "polymorphic region of a gene". A polymorphic region can be a single nucleotide, the identity of which differs in different alleles. A polymorphic region can also be several nucleotides long.

10 A "polymorphic gene" refers to a gene having at least one polymorphic region.

To describe a "polymorphic site" in a nucleotide sequence often there is used an "ambiguity code" that stands for the possible variations of nucleotides in one site.

The list of ambiguity codes is summarized in the following table:

15

Ambiguity (IUPAC Nomenclature)	Codes
B	c/g/t
D	a/g/t
H	a/c/t
K	g/t
M	a/c
N	a/c/g/t
R	a/g
S	c/g
V	a/c/g
W	a/t
Y	c/t

So, for example, a "R" in a nucleotide sequence means that either an "a" or a "g" could be at that position.

The terms "protein", "polypeptide" and "peptide" are used interchangeably herein when referring to a gene product.

- 5 A "regulatory element", also termed herein "regulatory sequence is intended to include elements which are capable of modulating transcription from a basic promoter and include elements such as enhancers and silencers. The term "enhancer", also referred to herein as "enhancer element", is intended to include regulatory elements capable of increasing, stimulating, or enhancing transcription from a basic 10 promoter. The term "silencer", also referred to herein as "silencer element" is intended to include regulatory elements capable of decreasing, inhibiting, or repressing transcription from a basic promoter. Regulatory elements are typically present in 5' flanking regions of genes. However, regulatory elements have also been shown to be present in other regions of a gene, in particular in introns. Thus, it is 15 possible that genes have regulatory elements located in introns, exons, coding regions, and 3' flanking sequences. Such regulatory elements are also intended to be encompassed by the present invention and can be identified by any of the assays that can be used to identify regulatory elements in 5' flanking regions of genes.
- 20 The term "regulatory element" further encompasses "tissue specific" regulatory elements, i.e., regulatory elements which effect expression of the selected DNA sequence preferentially in specific cells (e.g., cells of a specific tissue). gene expression occurs preferentially in a specific cell if expression in this cell type is significantly higher than expression in other cell types. The term "regulatory 25 element", also encompasses non-tissue specific regulatory elements, i.e., regulatory elements which are active in most cell types. Furthermore, a regulatory element can be a constitutive regulatory element, i.e., a regulatory element which constitutively regulates transcription, as opposed to a regulatory element which is inducible, i.e., a regulatory element which is active primarily in response to a stimulus. A stimulus can be, e.g., a molecule, such as a hormone, cytokine, heavy metal, phorbol ester, 30 cyclic AMP (cAMP), or retinoic acid.

Regulatory elements are typically bound by proteins, e.g., transcription factors. The term "transcription factor" is intended to include proteins or modified forms thereof, which interact preferentially with specific nucleic acid sequences, i.e., regulatory elements, and which in appropriate conditions stimulate or repress transcription.

5 Some transcription factors are active when they are in the form of a monomer. Alternatively, other transcription factors are active in the form of a dimer consisting of two identical proteins or different proteins (heterodimer). Modified forms of transcription factors are intended to refer to transcription factors having a post-translational modification, such as the attachment of a phosphate group. The activity of a transcription factor is frequently modulated by a post-translational modification. For example, certain transcription factors are active only if they are phosphorylated on specific residues. Alternatively, transcription factors can be active in the absence of phosphorylated residues and become inactivated by phosphorylation. A list of known transcription factors and their DNA binding site can be found, e.g., in public databases, e.g., TFMATRIX Transcription Factor Binding Site Profile database.

As used herein, the term "specifically hybridizes" or "specifically detects" refers to the ability of a nucleic acid molecule of the invention to hybridize to at least approximately 6, 12, 20, 30, 40, 50, 60, 70, 80, 90, 100, 110, 120, 130 or 140 consecutive nucleotides of either strand of a gene.

The term "wild-type allele" refers to an allele of a gene which, when present in two copies in a subject results in a wild-type phenotype. There can be several different wild-type alleles of a specific gene, since certain nucleotide changes in a gene may not affect the phenotype of a subject having two copies of the gene with the nucleotide changes.

"Adverse drug reaction" (ADR) as used herein refers to an appreciably harmful or unpleasant reaction, resulting from an intervention related to the use of a medicinal product, which predicts hazard from future administration and warrants prevention or

specific treatment, or alteration of the dosage regimen, or withdrawal of the product. In it's most severe form an ADR might lead to the death of an individual.

The term "Drug Response" is intended to mean any response that a patient exhibits upon drug administration. Specifically drug response includes beneficial, i.e. desired drug effects, ADR or no detectable reaction at all. More specifically the term drug response could also have a qualitative meaning, i.e. it embraces low or high beneficial effects, respectively and mild or severe ADR, respectively. The term "Statin Response" as used herein refers to drug response after statin administration.

An individual drug response includes also a good or bad metabolizing of the drug, meaning that "bad metabolizers" accumulate the drug in the body and by this could show side effects of the drug due to accumulative overdoses.

"Candidate gene" as used herein includes genes that can be assigned to either normal cardiovascular function or to metabolic pathways that are related to onset and/or progression of cardiovascular diseases.

With regard to drug response the term "candidate gene" includes genes that can be assigned to distinct phenotypes regarding the patient's response to drug administration. Those phenotypes may include patients who benefit from relatively small amounts of a given drug (high responders) or patients who need relatively high doses in order to obtain the same benefit (low responders). In addition those phenotypes may include patients who can tolerate high doses of a medicament without exhibiting ADR, or patients who suffer from ADR even after receiving only low doses of a medicament.

As neither the development of cardiovascular diseases nor the patient's response to drug administration is completely understood, the term "candidate gene" may also comprise genes with presently unknown function.

"PA SNP" (phenotype associated SNP) refers to a polymorphic site which shows a significant association with a patients phenotype (healthy, diseased, low or high responder, drug tolerant, ADR prone, etc.)

- 5 "PA gene" (phenotype associated gene) refers to a genomic locus harbouring a PA SNP, irrespective of the actual function of this gene locus.

PA gene polypeptide refers to a polypeptide encoded at least in part by a PA gene.

- 10 The term "Haplotype" as used herein refers to a group of two or more SNPs that are functionally and/or spatially linked. I.e. haplotypes define groups of SNPs that lie inside genes belonging to identical (or related metabolic) pathways and/or lie on the same chromosome. Haplotypes are expected to give better predictive/diagnostic information than a single SNP

- 15 The term "statin" is intended to embrace all inhibitors of the enzyme 3-hydroxy-3-methylglutaryl coenzyme A (HMG-CoA) reductase. Statins specifically inhibit the enzyme HMG-CoA reductase which catalyzes the rate limiting step in cholesterol biosynthesis. Known statins are Atorvastatin, Cerivastatin, Fluvastatin, Lovastatin, 20 Pravastatin and Simvastatin.

Methods for Assessing Cardiovascular Status

- The present invention provides diagnostic methods for assessing cardiovascular status in a human individual. Cardiovascular status as used herein refers to the physiological status of an individual's cardiovascular system as reflected in one or more markers or indicators. Status markers include without limitation clinical measurements such as, e.g., blood pressure, electrocardiographic profile, and differentiated blood flow analysis as well as measurements of LDL- and HDL-Cholesterol levels, other lipids and other well established clinical parameters that are standard in the art. Status markers according to the invention include diagnoses of 25 30

one or more cardiovascular syndromes, such as, e.g., hypertension, acute myocardial infarction, silent myocardial infarction, stroke, and atherosclerosis. It will be understood that a diagnosis of a cardiovascular syndrome made by a medical practitioner encompasses clinical measurements and medical judgement. Status markers according to the invention are assessed using conventional methods well known in the art. Also included in the evaluation of cardiovascular status are quantitative or qualitative changes in status markers with time, such as would be used, e.g., in the determination of an individual's response to a particular therapeutic regimen.

10

The methods are carried out by the steps of:

(i) determining the sequence of one or more polymorphic positions within one, several or all of the genes listed in Examples or other genes mentioned in this file in the individual to establish a polymorphic pattern for the individual; and

(ii) comparing the polymorphic pattern established in (i) with the polymorphic patterns of humans exhibiting different markers of cardiovascular status. The polymorphic pattern of the individual is, preferably, highly similar and, most preferably, identical to the polymorphic pattern of individuals who exhibit particular status markers, cardiovascular syndromes, and/or particular patterns of response to therapeutic interventions. Polymorphic patterns may also include polymorphic positions in other genes which are shown, in combination with one or more polymorphic positions in the genes listed in the Examples, to correlate with the presence of particular status markers. In one embodiment, the method involves comparing an individual's polymorphic pattern with polymorphic patterns of individuals who have been shown to respond positively or negatively to a particular therapeutic regimen. Therapeutic regimen as used herein refers to treatments aimed at the elimination or amelioration of symptoms and events associated cardiovascular disease. Such treatments include without limitation one or more of alteration in diet, lifestyle, and exercise regimen; invasive and noninvasive surgical techniques such as

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atherectomy, angioplasty, and coronary bypass surgery; and pharmaceutical interventions, such as administration of ACE inhibitors, angiotensin II receptor antagonists, diuretics, alpha-adrenoreceptor antagonists, cardiac glycosides, phosphodiesterase inhibitors, beta-adrenoreceptor antagonists, calcium channel blockers, HMG-CoA reductase inhibitors, imidazoline receptor blockers, endothelin receptor blockers, organic nitrates, and modulators of protein function of genes listed in the Examples. Interventions with pharmaceutical agents not yet known whose activity correlates with particular polymorphic patterns associated with cardiovascular disease are also encompassed. It is contemplated, for example, that patients who are candidates for a particular therapeutic regimen will be screened for polymorphic patterns that correlate with responsivity to that particular regimen.

In a preferred embodiment, the method involves comparing an individual's polymorphic pattern with polymorphic patterns of individuals who exhibit or have exhibited one or more markers of cardiovascular disease, such as, e.g., elevated LDL-Cholesterol levels, high blood pressure, abnormal electrocardiographic profile, myocardial infarction, stroke, or atherosclerosis.

In another embodiment, the method involves comparing an individual's polymorphic pattern with polymorphic patterns of individuals who exhibit or have exhibited one or more drug related phenotypes, such as, e.g., low or high drug response, or adverse drug reactions.

In practicing the methods of the invention, an individual's polymorphic pattern can be established by obtaining DNA from the individual and determining the sequence at predetermined polymorphic positions in the genes such as those described in this file.

The DNA may be obtained from any cell source. Non-limiting examples of cell sources available in clinical practice include blood cells, buccal cells, cervicovaginal cells, epithelial cells from urine, fetal cells, or any cells present in tissue obtained by

biopsy. Cells may also be obtained from body fluids, including without limitation blood, saliva, sweat, urine, cerebrospinal fluid, feces, and tissue exudates at the site of infection or inflammation. DNA is extracted from the cell source or body fluid using any of the numerous methods that are standard in the art. It will be understood
5 that the particular method used to extract DNA will depend on the nature of the source.

Diagnostic and Prognostic Assays

10 The present invention provides methods for determining the molecular structure of at least one polymorphic region of a gene, specific allelic variants of said polymorphic region being associated with cardiovascular disease. In one embodiment, determining the molecular structure of a polymorphic region of a gene comprises determining the identity of the allelic variant. A polymorphic region of a gene, of which specific
15 alleles are associated with cardiovascular disease can be located in an exon, an intron, at an intron/exon border, or in the promoter of the gene.

The invention provides methods for determining whether a subject has, or is at risk, of developing a cardiovascular disease. Such disorders can be associated with an
20 aberrant gene activity, e.g., abnormal binding to a form of a lipid, or an aberrant gene protein level. An aberrant gene protein level can result from an aberrant transcription or post-transcriptional regulation. Thus, allelic differences in specific regions of a gene can result in differences of gene protein due to differences in regulation of expression. In particular, some of the identified polymorphisms in the human gene
25 may be associated with differences in the level of transcription; RNA maturation; splicing, or translation of the gene or transcription product.

In preferred embodiments, the methods of the invention can be characterized as comprising detecting, in a sample of cells from the subject, the presence or absence
30 of a specific allelic variant of one or more polymorphic regions of a gene. The allelic differences can be: (i) a difference in the identity of at least one nucleotide or (ii) a

difference in the number of nucleotides, which difference can be a single nucleotide or several nucleotides.

A preferred detection method is allele specific hybridization using probes overlapping the polymorphic site and having about 5, 10, 20, 25, or 30 nucleotides around the polymorphic region. Examples of probes for detecting specific allelic variants of the polymorphic region located in intron X are probes comprising a nucleotide sequence set forth in any of SEQ ID NO. X. In a preferred embodiment of the invention, several probes capable of hybridizing specifically to allelic variants are attached to a solid phase support, e.g., a "chip". Oligonucleotides can be bound to a solid support by a variety of processes, including lithography. For example a chip can hold up to 250,000 oligonucleotides (GeneChip, Affymetrix). Mutation detection analysis using these chips comprising oligonucleotides, also termed "DNA probe arrays" is described e.g., in Cronin et al. (1996) Human Mutation 7:244 and in Kozal et al. (1996) Nature Medicine 2:753. In one embodiment, a chip comprises all the allelic variants of at least one polymorphic region of a gene. The solid phase support is then contacted with a test nucleic acid and hybridization to the specific probes is detected. Accordingly, the identity of numerous allelic variants of one or more genes can be identified in a simple hybridization experiment. For example, the identity of the allelic variant of the nucleotide polymorphism of nucleotide A or G at position 33 of Seq ID 1 (baySNP179) and that of other possible polymorphic regions can be determined in a single hybridization experiment.

In other detection methods, it is necessary to first amplify at least a portion of a gene prior to identifying the allelic variant. Amplification can be performed, e.g., by PCR and/or LCR, according to methods known in the art. In one embodiment, genomic DNA of a cell is exposed to two PCR primers and amplification for a number of cycles sufficient to produce the required amount of amplified DNA. In preferred embodiments, the primers are located between 40 and 350 base pairs apart. Preferred primers for amplifying gene fragments of genes of this file are listed in Table 2 in the Examples.

Alternative amplification methods include: self sustained sequence replication (Guatelli, J. C. et al., 1990, Proc. Natl. Acad. Sci. U.S.A. 87:1874-1878), transcriptional amplification system (Kwoh, D. Y. et al., 1989, Proc. Natl. Acad. Sci. U.S.A. 86:1173-1177), Q-Beta Replicase (Lizardi, P. M. et al., 1988, Bio/Technology 6:1197), or any other nucleic acid amplification method, followed by the detection of the amplified molecules using techniques well known to those of skill in the art. These detection schemes are especially useful for the detection of nucleic acid molecules if such molecules are present in very low numbers.

10

In one embodiment, any of a variety of sequencing reactions known in the art can be used to directly sequence at least a portion of a gene and detect allelic variants, e.g., mutations, by comparing the sequence of the sample sequence with the corresponding wild-type (control) sequence. Exemplary sequencing reactions include those based on techniques developed by Maxam and Gilbert (Proc. Natl Acad Sci USA (1977) 74:560) or Sanger (Sanger et al (1977) Proc. Nat. Acad. Sci 74:5463). It is also contemplated that any of a variety of automated sequencing procedures may be utilized when performing the subject assays (Biotechniques (1995) 19:448), including sequencing by mass spectrometry (see, for example, U.S. Pat. No. 5,547,835 and international patent application Publication Number WO 94/16101, entitled DNA Sequencing by Mass Spectrometry by H. Koster; U.S. Pat. No. 5,547,835 and international patent application Publication Number WO 94/21822 entitled "DNA Sequencing by Mass Spectrometry Via Exonuclease Degradation" by H. Koster), and U.S. Pat. No. 5,605,798 and International Patent Application No. PCT/US96/03651 entitled DNA Diagnostics Based on Mass Spectrometry by H. Koster; Cohen et al. (1996) Adv Chromatogr 36:127-162; and Griffin et al. (1993) Appl Biochem Biotechnol 38:147-159). It will be evident to one skilled in the art that, for certain embodiments, the occurrence of only one, two or three of the nucleic acid bases need be determined in the sequencing reaction. For instance, A-track or the like, e.g., where only one nucleotide is detected, can be carried out.

30

Yet other sequencing methods are disclosed, e.g., in U.S. Pat. No. 5,580,732 entitled "Method of DNA sequencing employing a mixed DNA-polymer chain probe" and U.S. Pat. No. 5,571,676 entitled "Method for mismatch-directed in vitro DNA sequencing".

5

In some cases, the presence of a specific allele of a gene in DNA from a subject can be shown by restriction enzyme analysis. For example, a specific nucleotide polymorphism can result in a nucleotide sequence comprising a restriction site which is absent from the nucleotide sequence of another allelic variant.

10

In other embodiments, alterations in electrophoretic mobility is used to identify the type of gene allelic variant. For example, single strand conformation polymorphism (SSCP) may be used to detect differences in electrophoretic mobility between mutant and wild type nucleic acids (Orita et al. (1989) Proc Natl. Acad. Sci USA 86:2766, see also Cotton (1993) Mutat Res 285:125-144; and Hayashi (1992) Genet Anal Tech Appl 9:73-79). Single-stranded DNA fragments of sample and control nucleic acids are denatured and allowed to renature. The secondary structure of single-stranded nucleic acids varies according to sequence, the resulting alteration in electrophoretic mobility enables the detection of even a single base change. The DNA fragments may be labeled or detected with labeled probes. The sensitivity of the assay may be enhanced by using RNA (rather than DNA), in which the secondary structure is more sensitive to a change in sequence. In another preferred embodiment, the subject method utilizes heteroduplex analysis to separate double stranded heteroduplex molecules on the basis of changes in electrophoretic mobility (Keen et al. (1991) Trends Genet 7:5).

In yet another embodiment, the identity of an allelic variant of a polymorphic region is obtained by analyzing the movement of a nucleic acid comprising the polymorphic region in polyacrylamide gels containing a gradient of denaturant is assayed using denaturing gradient gel electrophoresis (DGGE) (Myers et al (1985) Nature 313:495). When DGGE is used as the method of analysis, DNA will be modified to

insure that it does not completely denature, for example by adding a GC clamp of approximately 40 bp of high-melting GC-rich DNA by PCR. In a further embodiment, a temperature gradient is used in place of a denaturing agent gradient to identify differences in the mobility of control and sample DNA (Rosenbaum and
5 Reissner (1987) Biophys Chem 265:1275).

Examples of techniques for detecting differences of at least one nucleotide between 2 nucleic acids include, but are not limited to, selective oligonucleotide hybridization, selective amplification, or selective primer extension. For example, oligonucleotide probes may be prepared in which the known polymorphic nucleotide is placed centrally (allele-specific probes) and then hybridized to target DNA under conditions which permit hybridization only if a perfect match is found (Saiki et al. (1986) Nature 324:163); Saiki et al (1989) Proc. Natl Acad. Sci USA 86:6230; and Wallace et al. (1979) Nucl. Acids Res. 6:3543). Such allele specific oligonucleotide hybridization techniques may be used for the simultaneous detection of several nucleotide changes in different polymorphic regions of gene. For example, oligonucleotides having nucleotide sequences of specific allelic variants are attached to a hybridizing membrane and this membrane is then hybridized with labeled sample nucleic acid. Analysis of the hybridization signal will then reveal the identity of the nucleotides of
10 the sample nucleic acid.
15
20

Alternatively, allele specific amplification technology which depends on selective PCR amplification may be used. Oligonucleotides used as primers for specific amplification may carry the allelic variant of interest in the center of the molecule (so that amplification depends on differential hybridization) (Gibbs et al (1989) Nucleic Acids Res. 17:2437-2448) or at the extreme 3' end of one primer where, under appropriate conditions, mismatch can prevent, or reduce polymerase extension (Prossner (1993) Tibtech 11:238; Newton et al. (1989) Nucl. Acids Res. 17:2503). This technique is also termed "PROBE" for Probe Oligo Base Extension. In addition
25 it may be desirable to introduce a novel restriction site in the region of the mutation to create cleavage-based detection (Gasparini et al (1992) Mol. Cell Probes 6:1).
30

In another embodiment, identification of the allelic variant is carried out using an oligonucleotide ligation assay (OLA), as described, e.g., in U.S. Pat. No. 4,998,617 and in Landegren, U. et al., Science 241:1077-1080 (1988). The OLA protocol uses two oligonucleotides which are designed to be capable of hybridizing to abutting sequences of a single strand of a target. One of the oligonucleotides is linked to a separation marker, e.g., biotinylated, and the other is detectably labeled. If the precise complementary sequence is found in a target molecule, the oligonucleotides will hybridize such that their termini abut, and create a ligation substrate. Ligation then permits the labeled oligonucleotide to be recovered using avidin, or another biotin ligand. Nickerson, D. A. et al. have described a nucleic acid detection assay that combines attributes of PCR and OLA (Nickerson, D. A. et al., Proc. Natl. Acad. Sci. (U.S.A.) 87:8923-8927 (1990). In this method, PCR is used to achieve the exponential amplification of target DNA, which is then detected using OLA.

Several techniques based on this OLA method have been developed and can be used to detect specific allelic variants of a polymorphic region of a gene. For example, U.S. Pat. No. 5,593,826 discloses an OLA using an oligonucleotide having 3'-amino group and a 5'-phosphorylated oligonucleotide to form a conjugate having a phosphoramidate linkage. In another variation of OLA described in Tobe et al. ((1996)Nucleic Acids Res 24: 3728), OLA combined with PCR permits typing of two alleles in a single microtiter well. By marking each of the allele-specific primers with a unique hapten, i.e. digoxigenin and fluorescein, each LA reaction can be detected by using hapten specific antibodies that are labeled with different enzyme reporters, alkaline phosphatase or horseradish peroxidase. This system permits the detection of the two alleles using a high throughput format that leads to the production of two different colors.

The invention further provides methods for detecting single nucleotide polymorphisms in a gene. Because single nucleotide polymorphisms constitute sites of variation flanked by regions of invariant sequence, their analysis requires no more

than the determination of the identity of the single nucleotide present at the site of variation and it is unnecessary to determine a complete gene sequence for each patient. Several methods have been developed to facilitate the analysis of such single nucleotide polymorphisms.

5

In one embodiment, the single base polymorphism can be detected by using a specialized exonuclease-resistant nucleotide, as disclosed, e.g., in Mundy, C. R. (U.S. Pat. No. 4,656,127). According to the method, a primer complementary to the allelic sequence immediately 3^t to the polymorphic site is permitted to hybridize to a target 10 molecule obtained from a particular animal or human. If the polymorphic site on the target molecule contains a nucleotide that is complementary to the particular exonuclease-resistant nucleotide derivative present, then that derivative will be incorporated onto the end of the hybridized primer. Such incorporation renders the primer resistant to exonuclease, and thereby permits its detection. Since the identity 15 of the exonuclease-resistant derivative of the sample is known, a finding that the primer has become resistant to exonucleases reveals that the nucleotide present in the polymorphic site of the target molecule was complementary to that of the nucleotide derivative used in the reaction. This method has the advantage that it does not require the determination of large amounts of extraneous sequence data.

20

In another embodiment of the invention, a solution-based method is used for determining the identity of the nucleotide of a polymorphic site. Cohen, D. et al. (French Patent 2,650,840; PCT Appln. No. WO91/02087). As in the Mundy method of U.S. Pat. No. 4,656,127, a primer is employed that is complementary to allelic 25 sequences immediately 3^t to a polymorphic site. The method determines the identity of the nucleotide of that site using labeled dideoxynucleotide derivatives, which, if complementary to the nucleotide of the polymorphic site will become incorporated onto the terminus of the primer.

30

An alternative method, known as Genetic Bit Analysis or GBA TM is described by Goelet, P. et al. (PCT Appln. No. 92/15712). The method of Goelet, P. et al. uses

mixtures of labeled terminators and a primer that is complementary to the sequence 3' to a polymorphic site. The labeled terminator that is incorporated is thus determined by, and complementary to, the nucleotide present in the polymorphic site of the target molecule being evaluated. In contrast to the method of Cohen et al. 5 (French Patent 2,650,840; PCT Appln. No. WO91/02087) the method of Goelet, P. et al. is preferably a heterogeneous phase assay, in which the primer or the target molecule is immobilized to a solid phase.

Recently, several primer-guided nucleotide incorporation procedures for assaying 10 polymorphic sites in DNA have been described (Komher, J. S. et al., Nucl. Acids Res. 17:7779-7784 (1989); Sokolov, B. P., Nucl. Acids Res. 18:3671 (1990); Syvanen, A.-C., et al., Genomics 8:684-692 (1990), Kuppuswamy, M. N. et al., Proc. Natl. Acad. Sci. (U.S.A.) 88:1143-1147 (1991); Prezant, T. R. et al., Hum. Mutat. 1:159-164 (1992); Uguzzoli, L. et al., GATA 9:107-112 (1992); Nyren, P. et 15 al., Anal. Biochem. 208:171-175 (1993)). These methods differ from GBA TM in that they all rely on the incorporation of labeled deoxynucleotides to discriminate between bases at a polymorphic site. In such a format, since the signal is proportional to the number of deoxynucleotides incorporated, polymorphisms that occur in runs of the same nucleotide can result in signals that are proportional to the length of the run. 20 (Syvanen, A.-C., et al., Amer. J. Hum. Genet. 52:46-59 (1993)).

For determining the identity of the allelic variant of a polymorphic region located in 25 the coding region of a gene, yet other methods than those described above can be used. For example, identification of an allelic variant which encodes a mutated gene protein can be performed by using an antibody specifically recognizing the mutant protein in, e.g., immunohistochemistry or immunoprecipitation. Antibodies to wild-type gene protein are described, e.g., in Acton et al. (1999) Science 271:518 (anti-mouse gene antibody cross-reactive with human gene). Other antibodies to wild-type gene or mutated forms of gene proteins can be prepared according to methods known 30 in the art. Alternatively, one can also measure an activity of a gene protein, such as binding to a lipid or lipoprotein. Binding assays are known in the art and involve,

e.g., obtaining cells from a subject, and performing binding experiments with a labeled lipid, to determine whether binding to the mutated form of the receptor differs from binding to the wild-type of the receptor.

5 If a polymorphic region is located in an exon, either in a coding or non-coding region of the gene, the identity of the allelic variant can be determined by determining the molecular structure of the mRNA, pre-mRNA, or cDNA. The molecular structure can be determined using any of the above described methods for determining the molecular structure of the genomic DNA, e.g., sequencing and SSCP.

10

The methods described herein may be performed, for example, by utilizing pre-packaged diagnostic kits, such as those described above, comprising at least one probe or primer nucleic acid described herein, which may be conveniently used, e.g., to determine whether a subject has or is at risk of developing a disease associated with a specific gene allelic variant.

15

Sample nucleic acid for using in the above-described diagnostic and prognostic methods can be obtained from any cell type or tissue of a subject. For example, a subject's bodily fluid (e.g. blood) can be obtained by known techniques (e.g. venipuncture) or from human tissues like heart (biopsies, transplanted organs). Alternatively, nucleic acid tests can be performed on dry samples (e.g. hair or skin). Fetal nucleic acid samples for prenatal diagnostics can be obtained from maternal blood as described in International Patent Application No. WO91/07660 to Bianchi. Alternatively, amniocytes or chorionic villi may be obtained for performing prenatal testing.

25

Diagnostic procedures may also be performed *in situ* directly upon tissue sections (fixed and/or frozen) of patient tissue obtained from biopsies or resections, such that no nucleic acid purification is necessary. Nucleic acid reagents may be used as probes and/or primers for such *in situ* procedures (see, for example, Nuovo, G. J.,

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1992, PCR in situ hybridization: protocols and applications, Raven Press, New York).

In addition to methods which focus primarily on the detection of one nucleic acid
5 sequence, profiles may also be assessed in such detection schemes. Fingerprint profiles may be generated, for example, by utilizing a differential display procedure, Northern analysis and/or RT-PCR.

In practicing the present invention, the distribution of polymorphic patterns in a large
10 number of individuals exhibiting particular markers of cardiovascular status or drug response is determined by any of the methods described above, and compared with the distribution of polymorphic patterns in patients that have been matched for age, ethnic origin, and/or any other statistically or medically relevant parameters, who exhibit quantitatively or qualitatively different status markers. Correlations are
15 achieved using any method known in the art, including nominal logistic regression, chi square tests or standard least squares regression analysis. In this manner, it is possible to establish statistically significant correlations between particular polymorphic patterns and particular cardiovascular statuses (given in p values). It is further possible to establish statistically significant correlations between particular
20 polymorphic patterns and changes in cardiovascular status or drug response such as, would result, e.g., from particular treatment regimens. In this manner, it is possible to correlate polymorphic patterns with responsivity to particular treatments.

In another embodiment of the present invention two or more polymorphic regions are
25 combined to define so called 'haplotypes'. Haplotypes are groups of two or more SNPs that are functionally and/or spatially linked. It is possible to combine SNPs that are disclosed in the present invention either with each other or with additional polymorphic regions to form a haplotype. Haplotypes are expected to give better predictive/diagnostic information than a single SNP.

30 In a preferred embodiment of the present invention a panel of SNPs/haplotypes is defined that predicts the risk for CVD or drug response. This predictive panel is then

used for genotyping of patients on a platform that can genotype multiple SNPs at the same time (Multiplexing). Preferred platforms are e.g. gene chips (Affymetrix) or the Luminex LabMAP reader. The subsequent identification and evaluation of a patient's haplotype can then help to guide specific and individualized therapy.

5

For example the present invention can identify patients exhibiting genetic polymorphisms or haplotypes which indicate an increased risk for adverse drug reactions. In that case the drug dose should be lowered in a way that the risk for ADR is diminished. Also if the patient's response to drug administration is particularly high (or the patient is badly metabolizing the drug), the drug dose should be lowered to avoid the risk of ADR.

In turn if the patient's response to drug administration is low (or the patient is a particularly high metabolizer of the drug), and there is no evident risk of ADR, the drug dose should be raised to an efficacious level.

15 It is self evident that the ability to predict a patient's individual drug response should affect the formulation of a drug, i.e. drug formulations should be tailored in a way that they suit the different patient classes (low/high responder, poor/good metabolizer, ADR prone patients). Those different drug formulations may encompass different doses of the drug, i.e. the medicinal products contains low or high amounts 20 of the active substance. In another embodiment of the invention the drug formulation may contain additional substances that facilitate the beneficial effects and/or diminish the risk for ADR (Folkers et al. 1991, US Pat. 5,316,765).

Isolated Polymorphic Nucleic Acids, Probes, and Vectors

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The present invention provides isolated nucleic acids comprising the polymorphic positions described herein for human genes; vectors comprising the nucleic acids; and transformed host cells comprising the vectors. The invention also provides probes which are useful for detecting these polymorphisms.

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In practicing the present invention, many conventional techniques in molecular biology, microbiology, and recombinant DNA, are used. Such techniques are well known and are explained fully in, for example, Sambrook et al., 1989, Molecular Cloning: A Laboratory Manual, Second Edition, Cold Spring Harbor Laboratory 5 Press, Cold Spring Harbor, New York; DNA Cloning: A Practical Approach, Volumes I and II, 1985 (D. N. Glover ed.); Oligonucleotide Synthesis, 1984, (M. L. Gait ed.); Nucleic Acid Hybridization, 1985, (Hames and Higgins); Ausubel et al., Current Protocols in Molecular Biology, 1997, (John Wiley and Sons); and Methods 10 in Enzymology Vol. 154 and Vol. 155 (Wu and Grossman, and Wu, eds., respectively).

Insertion of nucleic acids (typically DNAs) comprising the sequences in a functional surrounding like full length cDNA of the present invention into a vector is easily accomplished when the termini of both the DNAs and the vector comprise compatible restriction sites. If this cannot be done, it may be necessary to modify the 15 termini of the DNAs and/or vector by digesting back single-stranded DNA overhangs generated by restriction endonuclease cleavage to produce blunt ends, or to achieve the same result by filling in the single-stranded termini with an appropriate DNA polymerase.

Alternatively, any site desired may be produced, e.g., by ligating nucleotide 20 sequences (linkers) onto the termini. Such linkers may comprise specific oligonucleotide sequences that define desired restriction sites. Restriction sites can also be generated by the use of the polymerase chain reaction (PCR). See, e.g., Saiki et al., 1988, Science 239:48. The cleaved vector and the DNA fragments may also be 25 modified if required by homopolymeric tailing.

The nucleic acids may be isolated directly from cells or may be chemically 30 synthesized using known methods. Alternatively, the polymerase chain reaction (PCR) method can be used to produce the nucleic acids of the invention, using either chemically synthesized strands or genomic material as templates. Primers used for

PCR can be synthesized using the sequence information provided herein and can further be designed to introduce appropriate new restriction sites, if desirable, to facilitate incorporation into a given vector for recombinant expression.

- 5 The nucleic acids of the present invention may be flanked by native gene sequences, or may be associated with heterologous sequences, including promoters, enhancers, response elements, signal sequences, polyadenylation sequences, introns, 5'- and 3'- noncoding regions, and the like. The nucleic acids may also be modified by many means known in the art. Non-limiting examples of such modifications include
- 10 methylation, "caps", substitution of one or more of the naturally occurring nucleotides with an analog, internucleotide modifications such as, for example, those with uncharged linkages (e.g., methyl phosphonates, phosphotriesters, phosphoramidates, carbamates, morpholines etc.) and with charged linkages (e.g., phosphorothioates, phosphorodithioates, etc.). Nucleic acids may contain one or
- 15 more additional covalently linked moieties, such as, for example, proteins (e.g., nucleases, toxins, antibodies, signal peptides, poly-L-lysine, etc.), intercalators (e.g., acridine, psoralen, etc.), chelators (e.g., metals, radioactive metals, iron, oxidative metals, etc.), and alkylators. PNAs are also included. The nucleic acid may be derivatized by formation of a methyl or ethyl phosphotriester or an alkyl phosphoramidate linkage. Furthermore, the nucleic acid sequences of the present
- 20 invention may also be modified with a label capable of providing a detectable signal, either directly or indirectly. Exemplary labels include radioisotopes, fluorescent molecules, biotin; and the like.
- 25 The invention also provides nucleic acid vectors comprising the gene sequences or derivatives or fragments thereof of genes described in the Examples. A large number of vectors, including plasmid and fungal vectors, have been described for replication and/or expression in a variety of eukaryotic and prokaryotic hosts, and may be used for gene therapy as well as for simple cloning or protein expression. Non-limiting examples of suitable vectors include without limitation pUC plasmids, pET plasmids (Novagen, Inc., Madison, Wis.), or pRSET or pREP (Invitrogen, San Diego, Calif.),

and many appropriate host cells, using methods disclosed or cited herein or otherwise known to those skilled in the relevant art. The particular choice of vector/host is not critical to the practice of the invention.

5 Suitable host cells may be transformed/transfected/infected as appropriate by any suitable method including electroporation, CaCl_2 mediated DNA uptake, fungal or viral infection, microinjection, microprojectile, or other established methods. Appropriate host cells included bacteria, archebacteria, fungi, especially yeast, and plant and animal cells, especially mammalian cells. A large number of transcription
10 initiation and termination regulatory regions have been isolated and shown to be effective in the transcription and translation of heterologous proteins in the various hosts. Examples of these regions, methods of isolation, manner of manipulation, etc. are known in the art. Under appropriate expression conditions, host cells can be used as a source of recombinantly produced peptides and polypeptides encoded by genes
15 of the Examples. Nucleic acids encoding peptides or polypeptides from gene sequences of the Examples may also be introduced into cells by recombination events. For example, such a sequence can be introduced into a cell and thereby effect homologous recombination at the site of an endogenous gene or a sequence with substantial identity to the gene. Other recombination-based methods such as non-homologous recombinations or deletion of endogenous genes by homologous
20 recombination may also be used.

In case of proteins that form heterodimers or other multimers, both or all subunits have to be expressed in one system or cell.

25 The nucleic acids of the present invention find use as probes for the detection of genetic polymorphisms and as templates for the recombinant production of normal or variant peptides or polypeptides encoded by genes listed in the Examples.

30 Probes in accordance with the present invention comprise without limitation isolated nucleic acids of about 10-100 bp, preferably 15-75 bp and most preferably 17-25 bp

in length, which hybridize at high stringency to one or more of the polymorphic sequences disclosed herein or to a sequence immediately adjacent to a polymorphic position. Furthermore, in some embodiments a full-length gene sequence may be used as a probe. In one series of embodiments, the probes span the polymorphic positions in genes disclosed herein. In another series of embodiments, the probes correspond to sequences immediately adjacent to the polymorphic positions.

Polymorphic Polypeptides and Polymorphism-Specific Antibodies

10 The present invention encompasses isolated peptides and polypeptides encoded by genes listed in the Examples comprising polymorphic positions disclosed herein. In one preferred embodiment, the peptides and polypeptides are useful screening targets to identify cardiovascular drugs. In another preferred embodiments, the peptides and polypeptides are capable of eliciting antibodies in a suitable host animal that react specifically with a polypeptide comprising the polymorphic position and distinguish it from other polypeptides having a different sequence at that position.

20 Polypeptides according to the invention are preferably at least five or more residues in length, preferably at least fifteen residues. Methods for obtaining these polypeptides are described below. Many conventional techniques in protein biochemistry and immunology are used. Such techniques are well known and are explained in Immunochemical Methods in Cell and Molecular Biology, 1987 (Mayer and Waler, eds; Academic Press, London); Scopes, 1987, Protein Purification: Principles and Practice, Second Edition (Springer-Verlag, N.Y.) and Handbook of Experimental Immunology, 1986, Volumes I-IV (Weir and Blackwell eds.).

25 Nucleic acids comprising protein-coding sequences can be used to direct the ITT recombinant expression of polypeptides encoded by genes disclosed herein in intact cells or in cell-free translation systems. The known genetic code, tailored if desired for more efficient expression in a given host organism, can be used to synthesize oligonucleotides encoding the desired amino acid sequences. The polypeptides may

be isolated from human cells, or from heterologous organisms or cells (including, but not limited to, bacteria, fungi, insect, plant, and mammalian cells) into which an appropriate protein-coding sequence has been introduced and expressed. Furthermore, the polypeptides may be part of recombinant fusion proteins.

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Peptides and polypeptides may be chemically synthesized by commercially available automated procedures, including, without limitation, exclusive solid phase synthesis, partial solid phase methods, fragment condensation or classical solution synthesis. The polypeptides are preferably prepared by solid phase peptide synthesis as described by Merrifield, 1963, J. Am. Chem. Soc. 85:2149.

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Methods for polypeptide purification are well-known in the art, including, without limitation, preparative disc-gel electrophoresis, isoelectric focusing, HPLC, reversed-phase HPLC, gel filtration, ion exchange and partition chromatography, and countercurrent distribution. For some purposes, it is preferable to produce the polypeptide in a recombinant system in which the protein contains an additional sequence tag that facilitates purification, such as, but not limited to, a polyhistidine sequence. The polypeptide can then be purified from a crude lysate of the host cell by chromatography on an appropriate solid-phase matrix. Alternatively, antibodies produced against peptides encoded by genes disclosed herein, can be used as purification reagents. Other purification methods are possible.

15

The present invention also encompasses derivatives and homologues of the polypeptides. For some purposes, nucleic acid sequences encoding the peptides may be altered by substitutions, additions, or deletions that provide for functionally equivalent molecules, i.e., function-conservative variants. For example, one or more amino acid residues within the sequence can be substituted by another amino acid of similar properties, such as, for example, positively charged amino acids (arginine, lysine, and histidine); negatively charged amino acids (aspartate and glutamate); polar neutral amino acids; and non-polar amino acids.

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The isolated polypeptides may be modified by, for example, phosphorylation, sulfation, acylation, or other protein modifications. They may also be modified with a label capable of providing a detectable signal, either directly or indirectly, including, but not limited to, radioisotopes and fluorescent compounds.

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The present invention also encompasses antibodies that specifically recognize the polymorphic positions of the invention and distinguish a peptide or polypeptide containing a particular polymorphism from one that contains a different sequence at that position. Such polymorphic position-specific antibodies according to the present invention include polyclonal and monoclonal antibodies. The antibodies may be elicited in an animal host by immunization with peptides encoded by genes disclosed herein or may be formed by in vitro immunization of immune cells. The immuno-
10 genic components used to elicit the antibodies may be isolated from human cells or produced in recombinant systems. The antibodies may also be produced in
15 recombinant systems programmed with appropriate antibody-encoding DNA. Alternatively, the antibodies may be constructed by biochemical reconstitution of purified heavy and light chains. The antibodies include hybrid antibodies (i.e., containing two sets of heavy chain/light chain combinations, each of which recognizes a different antigen), chimeric antibodies (i.e., in which either the heavy
20 chains, light chains, or both, are fusion proteins), and univalent antibodies (i.e., comprised of a heavy chain/light chain complex bound to the constant region of a second heavy chain). Also included are Fab fragments, including Fab' and F(ab)₂ fragments of antibodies. Methods for the production of all of the above types of antibodies and derivatives are well-known in the art and are discussed in
25 more detail below. For example, techniques for producing and processing polyclonal antisera are disclosed in Mayer and Walker, 1987, Immunochemical Methods in Cell and Molecular Biology, (Academic Press, London). The general methodology for making monoclonal antibodies by hybridomas is well known. Immortal antibody-producing cell lines can be created by cell fusion, and also by other techniques such
30 as direct transformation of B lymphocytes with oncogenic DNA, or transfection with Epstein-Barr virus. See, e.g., Schreier et al., 1980, Hybridoma Techniques; U.S. Pat.

Nos. 4,341,761; 4,399,121; 4,427,783; 4,444,887; 4,466,917; 4,472,500; 4,491,632; and 4,493,890. Panels of monoclonal antibodies produced against peptides encoded by genes disclosed herein can be screened for various properties; i.e. for isotype, epitope affinity, etc.

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The antibodies of this invention can be purified by standard methods, including but not limited to preparative disc-gel electrophoresis, isoelectric focusing, HPLC, reversed-phase HPLC, gel filtration, ion exchange and partition chromatography, and countercurrent distribution. Purification methods for antibodies are disclosed, e.g., in

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The Art of Antibody Purification, 1989, Amicon Division, W. R. Grace & Co. General protein purification methods are described in Protein Purification: Principles and Practice, R. K. Scopes, Ed., 1987, Springer-Verlag, New York, N.Y.

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Methods for determining the immunogenic capability of the disclosed sequences and the characteristics of the resulting sequence-specific antibodies and immune cells are well-known in the art. For example, antibodies elicited in response to a peptide comprising a particular polymorphic sequence can be tested for their ability to specifically recognize that polymorphic sequence, i.e., to bind differentially to a peptide or polypeptide comprising the polymorphic sequence and thus distinguish it from a similar peptide or polypeptide containing a different sequence at the same position.

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Kits

As set forth herein, the invention provides diagnostic methods, e.g., for determining the identity of the allelic variants of polymorphic regions present in the gene loci of genes disclosed herein, wherein specific allelic variants of the polymorphic region are associated with cardiovascular diseases. In a preferred embodiment, the diagnostic kit can be used to determine whether a subject is at risk of developing a cardiovascular disease. This information could then be used, e.g., to optimize treatment of such individuals.

10:

In preferred embodiments, the kit comprises a probe or primer which is capable of hybridizing to a gene and thereby identifying whether the gene contains an allelic variant of a polymorphic region which is associated with a risk for cardiovascular disease. The kit preferably further comprises instructions for use in diagnosing a subject as having, or having a predisposition, towards developing a cardiovascular disease. The probe or primers of the kit can be any of the probes or primers described in this file.

Preferred kits for amplifying a region of a gene comprising a polymorphic region of interest comprise one, two or more primers.

Antibody-based diagnostic methods and kits:

The invention also provides antibody-based methods for detecting polymorphic patterns in a biological sample. The methods comprise the steps of: (i) contacting a sample with one or more antibody preparations, wherein each of the antibody preparations is specific for a particular polymorphic form of the proteins encoded by genes disclosed herein, under conditions in which a stable antigen-antibody complex can form between the antibody and antigenic components in the sample; and (ii) detecting any antigen-antibody complex formed in step (i) using any suitable means

known in the art, wherein the detection of a complex indicates the presence of the particular polymorphic form in the sample.

Typically, immunoassays use either a labelled antibody or a labelled antigenic component (e.g., that competes with the antigen in the sample for binding to the antibody). Suitable labels include without limitation enzyme-based, fluorescent, chemiluminescent, radioactive, or dye molecules. Assays that amplify the signals from the probe are also known, such as, for example, those that utilize biotin and avidin, and enzyme-labelled immunoassays, such as ELISA assays.

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The present invention also provides kits suitable for antibody-based diagnostic applications. Diagnostic kits typically include one or more of the following components:

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(i) Polymorphism-specific antibodies. The antibodies may be pre-labelled; alternatively, the antibody may be unlabelled and the ingredients for labelling may be included in the kit in separate containers, or a secondary, labelled antibody is provided; and

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(ii) Reaction components: The kit may also contain other suitably packaged reagents and materials needed for the particular immunoassay protocol, including solid-phase matrices, if applicable, and standards.

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The kits referred to above may include instructions for conducting the test. Furthermore, in preferred embodiments, the diagnostic kits are adaptable to high-throughput and/or automated operation.

Drug Targets and Screening Methods

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According to the present invention, nucleotide sequences derived from genes disclosed herein and peptide sequences encoded by genes disclosed herein, particularly

those that contain one or more polymorphic sequences, comprise useful targets to identify cardiovascular drugs, i.e., compounds that are effective in treating one or more clinical symptoms of cardiovascular disease. Furthermore, especially when a protein is a multimeric protein that are build of two or more subunits, is a combination of different polymorphic subunits very useful.

10 Drug targets include without limitation (i) isolated nucleic acids derived from the genes disclosed herein, and (ii) isolated peptides and polypeptides encoded by genes disclosed herein, each of which comprises one or more polymorphic positions.

15

In vitro screening methods:

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In one series of embodiments, an isolated nucleic acid comprising one or more polymorphic positions is tested in vitro for its ability to bind test compounds in a sequence-specific manner. The methods comprise:

(i) providing a first nucleic acid containing a particular sequence at a polymorphic position and a second nucleic acid whose sequence is identical to that of the first nucleic acid except for a different sequence at the same polymorphic position;

(ii) contacting the nucleic acids with a multiplicity of test compounds under conditions appropriate for binding; and

(iii) identifying those compounds that bind selectively to either the first or second nucleic acid sequence.

25 Selective binding as used herein refers to any measurable difference in any parameter of binding, such as, e.g., binding affinity, binding capacity, etc.

In another series of embodiments, an isolated peptide or polypeptide comprising one or more polymorphic positions is tested in vitro for its ability to bind test compounds in a sequence-specific manner. The screening methods involve:

- 5 (i) providing a first peptide or polypeptide containing a particular sequence at a polymorphic position and a second peptide or polypeptide whose sequence is identical to the first peptide or polypeptide except for a different sequence at the same polymorphic position;
- 10 (ii) contacting the polypeptides with a multiplicity of test compounds under conditions appropriate for binding; and
- 15 (iii) identifying those compounds that bind selectively to one of the nucleic acid sequences.

- 15 In preferred embodiments, high-throughput screening protocols are used to survey a large number of test compounds for their ability to bind the genes or peptides disclosed above in a sequence-specific manner.
- 20 Test compounds are screened from large libraries of synthetic or natural compounds. Numerous means are currently used for random and directed synthesis of saccharide, peptide, and nucleic acid based compounds. Synthetic compound libraries are commercially available from Maybridge Chemical Co. (Trevillet, Cornwall, UK), Comgenex (Princeton, N.J.), Brandon Associates (Merrimack, N.H.), and Microsource (New Milford, Conn.). A rare chemical library is available from Aldrich (Milwaukee, Wis.). Alternatively, libraries of natural compounds in the form of bacterial, fungal, plant and animal extracts are available from e.g. Pan Laboratories (Bothell, Wash.) or MycoSearch (N.C.), or are readily producible. Additionally, natural and synthetically produced libraries and compounds are readily modified through conventional chemical, physical, and biochemical means.
- 30

In vivo screening methods:

Intact cells or whole animals expressing polymorphic variants of genes disclosed herein can be used in screening methods to identify candidate cardiovascular drugs.

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In one series of embodiments, a permanent cell line is established from an individual exhibiting a particular polymorphic pattern. Alternatively, cells (including without limitation mammalian, insect, yeast, or bacterial cells) are programmed to express a gene comprising one or more polymorphic sequences by introduction of appropriate

10 DNA. Identification of candidate compounds can be achieved using any suitable assay, including without limitation (i) assays that measure selective binding of test compounds to particular polymorphic variants of proteins encoded by genes disclosed herein; (ii) assays that measure the ability of a test compound to modify (i.e., inhibit or enhance) a measurable activity or function of proteins encoded by
15 genes disclosed herein; and (iii) assays that measure the ability of a compound to modify (i.e., inhibit or enhance) the transcriptional activity of sequences derived from the promoter (i.e., regulatory) regions of genes disclosed herein.

20 In another series of embodiments, transgenic animals are created in which (i) one or more human genes disclosed herein, having different sequences at particular polymorphic positions are stably inserted into the genome of the transgenic animal; and/or (ii) the endogenous genes disclosed herein are inactivated and replaced with human genes disclosed herein, having different sequences at particular polymorphic positions. See, e.g., Coffman, Semin. Nephrol. 17:404, 1997; Esther et al., Lab.
25 Invest. 74:953, 1996; Murakami et al., Blood Press. Suppl. 2:36, 1996. Such animals can be treated with candidate compounds and monitored for one or more clinical markers of cardiovascular status.

The following are intended as non-limiting examples of the invention.

Material and Methods

Genotyping of patient DNA with the PyrosequencingTM Method as described in the patent application WO 9813523:

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First a PCR is set up to amplify the flanking regions around a SNP. Therefor 2 ng of genomic DNA (patient sample) are mixed with a primerset (20 – 40 pmol) producing a 75 to 320 bp PCR fragment with 0,3 to 1 U Qiagens Hot Star Taq PolymeraseTM in a total volume of 20 µL. One primer is biotinylated depending on the direction of the sequencing primer. To force the biotinylated primer to be incorporated it is used 0,8 fold.

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For primer design, programmes like Oligo 6TM (Molecular Biology Insights) or Primer SelectTM (DNAStar) are used. PCR setup is performed by a BioRobot 3000TM from Qiagen. PCR takes place in T1 or Tgradient ThermocyclersTM from Biometra. The whole PCR reaction is transferred into a PSQ plateTM (Pyrosequencing) and prepared using the Sample Prep ToolTM and SNP Reagent KitTM from Pyrosequencing according to their instructions.

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Preparation of template for PyrosequencingTM:

Sample preparation using PSQ 96 Sample Prep Tool:

1. Mount the PSQ 96 Sample Prep Tool Cover onto the PSQ 96 Sample Prep

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Tool as follows: Place the cover on the desk, retract the 4 attachment rods by separating the handle from the magnetic rod holder, fit the magnetic rods into the holes of the cover plate, push the handle downward until a click is heard.

The PSQ 96 Sample Prep Tool is now ready for use.

2. To transfer beads from one plate to another, place the covered tool into the

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PSQ 96 Plate containing the samples and lower the magnetic rods by separating the handle from the magnetic rod holder. Move the tool up and

down a few times then wait for 30-60 seconds. Transfer the beads into a new PSQ 96 plate containing the solution of choice.

3. Release the beads by lifting the magnetic rod holder, bringing it together with the handle. Move the tool up and down a few times to make sure that the beads are released.

5 All steps are performed at room temperature unless otherwise stated.

10 **Immobilization of PCR product:**

Biotinylated PCR products are immobilized on streptavidin-coated DynabeadsTM M-280 Streptavidin. Parallel immobilization of several samples are performed in the PSQ 96 Plate.

- 15 1. Mix PCR product, 20 µl of a well optimized PCR, with 25 µl 2X BW-buffer II. Add 60-150 µg Dynabeads: It is also possible to add a mix of Dynabeads and 2X BW-buffer II to the PCR product yielding a final BW-buffer II concentration of approximately 1x.

2. Incubate at 65°C for 15 min agitation constantly to keep the beads dispersed. 20 For optimal immobilization of fragments longer than 300 bp use 30 min incubation time.

Strand separation:

- 25 4. For strand separation, use the PSQ 96 Sample Prep Tool to transfer the beads with the immobilized sample to a PSQ 96 Plate containing 50 µl 0.50 M NaOH per well. Release the beads.

5. After approximately 1 min, transfer the beads with the immobilized strand to a PSQ 96 Plate containing 99 µl 1x Annealing buffer per well and mix thoroughly.

- 30 6. Transfer the beads to a PSQ 96 Plate containing 45 µl of a mix of 1x Annealing buffer and 3-15 pmoles sequencing primer per well.

7. Heat at 80°C for 2 minutes in the PSQ 96 Sample Prep Thermoplate and move to room temperature.
8. After reaching room temperature, continue with the sequencing reaction.

5

Sequencing reaction:

1. Choose the method to be used ("SNP Method") and enter relevant information in the PSQ 96 Instrument Control software.
2. Place the cartridge and PSQ 96 Plate in the PSQ 96 Instrument.
- 10 3. Start the run.

Genotyping using the ABI 7700/7900 instrument (TaqMan)

15 SNP genotyping using the TaqMan (Applied Biosystems/Perkin Elmer) was performed according to the manufacturer's instructions. The TaqMan assay is discussed by Lee et al., Nucleic Acids Research 1993, 21: 3761-3766.

20 **Genotyping with a service contractor:**

Qiagen Genomics, formerly Rapigene, is a service contractor for genotyping SNPs in patient samples. Their method is based on a primer extension method where two complementary primers are designed for each genotype that are labeled with different tags. Depending on the genotype only one primer will be elongated together with a certain tag. This tag can be detected with mass spectrometry and is a measure for the respective genotype. The method is described in the following patent: "Detection and identification of nucleic acid molecules - using tags which may be detected by non-fluorescent spectrometry or potentiometry" (WO 9727325).

Examples

To exemplify the present invention and its utility baySNP 28 will be used in the following:

5.

baySNP 28 is a C to T polymorphism and presumably resides in the gene of the human acidic 82 kDa protein (information taken from table 3). baySNP 28 was genotyped in various patient cohorts using the primers from table 2. As a result the following number of patients carrying different genotypes were found (information combined from tables 3 and 5a):

10.

baySNP	Cohort	Total	Genotype 11 "CC"	Genotype 12 "CT"	Genotype 22 "TT"
28	HELD_FEM_HIRESP	12	1	2	9
28	HELD_FEM_LORESP	22	3	12	7

When comparing the number of female patients exhibiting a high response to statin therapy (HELD_FEM_HIRESP) with the control cohort (HELD_FEM_LORESP) it appears that the number of low responders carrying the CT genotype is increased. This points to a lower statin response among female individuals with the CT genotype. Applying statistical tests on those findings the following p-values were obtained (data taken from table 5b):

15

20

BAYSNP	COMPARISON	GTYPE CPVAL	GTYPE XPVAL	GTYPE LRPVAL
28	HELD_FEM_EFF	0,0506	0,0508	0,0442

25

As at least one of the GTYPE p values is below 0,05 the association of genotype and statin response phenotype is regarded as statistically significant. I.e. the analysis of a patient's genotype can predict the response to statin therapy. In more detail one can calculate the relative risk to exhibit a certain statin response phenotype when carrying a certain genotype (data taken from table 6a):

BAYSNP	COMPARISON	GTYPE1	GTYPE2	GTYPE3	RR1	RR2	RR3
28	HELD_FEM_EFF	CC	CT	TT	0,68	0,29	3,38

In case of baySNP 28 the risk to exhibit a high responder phenotype is 3,38 times higher when carrying the TT genotype. This indicates that a TT polymorphism in baySNP 28 is an independent risk factor for high statin response in females. On the other hand carriers of a CT or CC genotype have a reduced risk of being a high responder.

In addition statistical associations can be calculated on the basis on alleles. This calculation would identify risk alleles instead of risk genotypes.

In case of baySNP 28 the following allele counts were obtained (data combined from tables 3 and 5a):

baySNP	Cohort	Total	Allele 1 "C"	Allele 2 "T"
28	HELD_FEM_HIRESP	12	4	20
28	HELD_FEM_LORESP	22	18	26

15

When comparing the number of female patients with high statin response (HELD_FEM_HIRESP) with the control cohort (HELD_FEM_LORESP) it appears that the number of high responders carrying the T allele is increased, whereas the number of high responders carrying the C allele is diminished. This points to a higher statin response among female individuals with the T allele. Applying statistical tests on those findings the following p-values were obtained (data taken from table 5b):

BAYSNP	COMPARISON	ALLELE CPVAL	ALLELE XPVAL	ALLELE LRPVAL
28	HELD_FEM_EFF	0,0411	0,0579	0,0349

As at least one of the ALLELE p values is below 0,05 the association of allele and statin response phenotype is regarded as statistically significant (in this example significant p values were obtained from two statistical tests). I.e. also the analysis of a patient's alleles from baySNP 28 can predict the extend of statin response. In more detail one can calculate the relative risk to exhibit a certain statin response phenotype when carrying a certain allele (data taken from table 6b):

baySNP	Allele 1	Allele 2	COMPARISON	RR1	RR2
28	C	T	HELD_FEM_EFF	0,42	2,39

- 10 In case of baySNP 28 the risk to exhibit a high responder phenotype is 2,39 times higher when carrying the T allele. This indicates that the T allele of baySNP28 is an independent risk factor for a high statin response in females. In other words those patients should receive lower doses of statins in order to avoid ADR. However due to their 'high responder' phenotype they will still benefit from the drug. In turn carriers of the C allele should receive higher drug doses in order to experience a benefical therapeutic effect.
- 15

- 20 Another example is baySNP 29, which is taken to exemplify polymorphisms relevant for adverse drug reactions. baySNP 29 was found significant when comparing male patients with severe ADR to the respective controls (as defined in table 1b).

The relative risk ratios for the genotypes AA, AG and GG were as follows (data taken from table 6a):

BAYSNP	COMPARISON	GTYPE1	GTYPE2	GTYPE3	RR1	RR2	RR3
29	HELD_MAL_ADR5ULN	AA	AG	GG	3,15	0,66	0,32

In this case male patients carrying the AA genotype have a 3,15 times higher risk to suffer from ADR. In other words those patients should either receive lower doses of statins or switch to an alternative therapy in order to avoid ADR. On the other hand male patients with AG or GG genotypes appear to be more resistant to ADR and hence better tolerate statin therapy.

5

As can be seen from the following tables some of the associations that are disclosed in the present invention are indicative for more than one phenotype. baySNP 1837 is for example linked to ADR, but also to the risk to suffer from CVD (table 6).

10

Table 1a Definition of "good" and "bad" serum lipid levels

	"Good"	"Bad"
LDL-Cholesterol [mg/dL]	125 -150	170 - 200
Cholesterol [mg/dL]	190 - 240	265 - 315
HDL-Cholesterol [mg/dL]	60 -105	30 - 55
Triglycerides [mg/dL]	45 - 115	170 – 450

Table 1b Definition of drug response phenotypes

Low responder	Decrease of serum LDL of at least 10% and at most 50% upon administration of 0.8 mg Cerivastatin (female patients)
High responder	Decrease of serum LDL of at least 50% upon administration of 0.4 mg Cerivastatin (female patients)
Very low responder	Decrease of serum LDL of at least 10% and at most 35% upon administration of 0.8 mg Cerivastatin (female patients)
Very high responder	Decrease of serum LDL of at least 55% upon administration of 0.4 mg Cerivastatin (female patients)

Ultra low responder	Decrease of serum LDL of at least 10% and at most 25% upon administration of 0.8 mg Cerivastatin (female patients)
Ultra high responder	Decrease of serum LDL of at least 60% upon administration of 0.4 mg Cerivastatin (female patients)
Tolerant patient	No diagnosis of muscle cramps, muscle pain, muscle weakness, myalgia or myopathy AND serum CK levels below 70 mg/dl in women and below 80 mg/dl in men.
ADR patient (CK increase at least 2×ULN)	Diagnosis of muscle cramps, muscle pain, muscle weakness, myalgia or myopathy OR serum CK levels higher than 140 mg/dl in women and 160 mg/dl in men.
Advanced ADR patient [ADR3] (advanced CK increase, at least 3×ULN)*	Serum CK levels higher than 210 mg/dl in women and 240 mg/dl in men
Severe ADR patient [ADR5] (severe CK increase, at least 5×ULN)*	Serum CK levels higher than 350 mg/dl in women and 400 mg/dl in men

*: When assembling the cohorts for advanced and severe ADR we focused on the CK serum levels as those provide a more independent measure of statin related ADR.

Table 1c Definition of "high" and "low" serum HDL cholesterol levels

	Male individuals	Female individuals
High* HDL-Cholesterol [mg/dL]	>=80	>=104
Low* HDL-Cholesterol [mg/dL]	<=35	<=37

5 An informed consent was signed by the patients and control people. Blood was taken by a physician according to medical standard procedures.

Samples were collected anonymous and labeled with a patient number.

DNA was extracted using kits from Qiagen.

Table 2a Oligonucleotide primers used for genotyping using mass spectrometry

10 The baySNP number refers to an internal numbering of the PA SNPs. Primer sequences are listed for preamplification of the genomic fragments (primers EF and ER) and for subsequent allele specific PCR of the SNP.

baySNP	SNP	Name	Sequence
28	C137T	CF	gggacggtcggtagatTCTAGAATTGTGCTTCCC
28	C137T	EF	TGTCCAGTGTTAGGAAAAA
28	C137T	ER	GACGATGCCTTCAGCACAGATGTGGCTTCTGTATGAG
28	C137T	TF	gctggctcggtcaagaTCTAGAATTGTGCTTCCT
29	A464G	AF	gggacggtcggtagatCATCGGTCA GTGTCCCCA
29	A464G	EF	GATGTC TGTCTCCTTGATGT
29	A464G	ER	GACGATGCCTTCAGCACAAATGTGGGGTTTATTT
29	A464G	GF	gctggctcggtcaagaCATCGGTCA GTGTCCCCG
52	C397G	CR	gggacggtcggtagatTATTTATAATGCAAAG
52	C397G	EF	GACGATGCCTTCAGCACAGTGAATTGCCAGATTAGTG
52	C397G	ER	TCTAAAGTGCTGGATTG
52	C397G	GR	gctggctcggtcaagaTATTTATAATGCAAAC
56	A429G	AF	gggacggtcggtagatAAGGTCTTGTACGTGTA

baySNP	SNP	Name	Sequence
56	A429G	EF	CCAGGTACTGCCTTACAAA
56	A429G	ER	GACGATGCCTTCAGCACAGCTCCAAAATAATCACTC
56	A429G	GF	gctggctcggtcaagaAAGGTCTTGTACGTGTG
89	A159G	AR	gggacggtcggtagatTGGAGTCGGGGAGTCAT
89	A159G	EF	GACGATGCCTTCAGCACATAGTTCAAGGGTAAAGGA
89	A159G	ER	GAGGACGAGATGTAAGAG
89	A159G	GR	gctggctcggtcaagaTGGAGTCGGGGAGTCAC
90	C154T	CF	gggacggtcggtagatCAGCGCATCCTGAACCAC
90	C154T	EF	GCTGGAACGAGTTCATCCT
90	C154T	ER	GACGATGCCTTCAGCACAGGACCCCACCTTCTTGT
90	C154T	TF	gctggctcggtcaagaCAGCGCATCCTGAACCAC
99	C58T	CR	gggacggtcggtagatTCCTGCTCTTCTCTAG
99	C58T	EF	GACGATGCCTTCAGCACACACTGACTGCTTACTCTACC
99	C58T	ER	TACTGTGTCTCAGCTCCA
99	C58T	TR	gctggctcggtcaagaTCCTGCTCTTCTCTAA
140	C468T	CR	gggacggtcggtagatGTGAATCCCAATACGAAG
140	C468T	EF	GACGATGCCTTCAGCACATAAAAATAACCAGGTACTCCA
140	C468T	ER	GATGAGTCCTCACCAAACATACA
140	C468T	TR	gctggctcggtcaagaGTGAATCCCAATACGAAA
152	A587G	AF	gggacggtcggtagatGGTGGGAGGTTCCAGCCA
152	A587G	EF	GCAGGAAGAAAGCTAGAA
152	A587G	ER	GACGATGCCTTCAGCACAAAGGCAGGATAATGACAAC
152	A587G	GF	gctggctcggtcaagaGGTGGGAGGTTCCAGCCG
214	A209G	AF	gggacggtcggtagatCATTCCACCTCACCAAA
214	A209G	EF	AGGTATTCCCGGCGTTTC
214	A209G	ER	GACGATGCCTTCAGCACATGTTGTGCGTCTGCTTCC
214	A209G	GF	gctggctcggtcaagaCATTCCACCTCACCAAG
221	C339G	CF	gggacggtcggtagatTGTGAAGAACTGTTGCTC
221	C339G	EF	CTGAAGCTCATCTGCCTTCT
221	C339G	ER	GACGATGCCTTCAGCACATCCCCCTCCTTACCT
221	C339G	GF	gctggctcggtcaagaTGTGAAGAACTGTTGCTG

baySNP	SNP	Name	Sequence
224	C189T	CR	gggacggtcggtagatGCCCGCTTTCTTCATCG
224	C189T	EF	GACGATGCCTTCAGCACACTGTCTCAAGGGCTTACAC
224	C189T	ER	TCCAACCTCAGGCAAAAC
224	C189T	TR	gctggctcggtcaagaGCCCGCTTTCTTCATCA
294	C465T	CR	gggacggtcggtagatCCCAAGGCCAACAGGGAG
294	C465T	EF	GACGATGCCTTCAGCACAGCATTCTATGCCAGTGTTC
294	C465T	ER	ATCCATCCCATCCTGTGT
294	C465T	TR	gctggctcggtcaagaCCCAAGGCCAACAGGGAA
307	C215T	CR	gggacggtcggtagatGAGTGGGTGCTGTTCCCG
307	C215T	EF	GACGATGCCTTCAGCACAGTTACTGCCTCTCTGACC
307	C215T	ER	AGTGTGACCTGCTCTCTT
307	C215T	TR	gctggctcggtcaagaGAGTGGGTGCTGTTCCCA
411	A369T	ER	gacgatgccttcagcacaAACACATTCCCCCTCTAC
411	A369T	EF	GTCTCTATTCCAAGCCAAG
411	A369T	AF	gggacggtcggtagatCCCGCTCCAGCTCCTCA
411	A369T	TF	gctggctcggtcaagaCCCGCTCCAGCTCCTCT
449	C323G	CR	gggacggtcggtagatCCGCTCTGCTTCTGCTG
449	C323G	EF	GACGATGCCTTCAGCACACAAGGAGAAGAGGGAGGAGA
449	C323G	ER	GGAGCACGTAAGGAGAAA
449	C323G	GR	gctggctcggtcaagaCCGCTCTGCTTCTGCTC
466	C123T	CF	gggacggtcggtagatGGCCAGGGGCTGGAGGGC
466	C123T	EF	TCTTCAGTTCTCTCAGCTTC
466	C123T	ER	GACGATGCCTTCAGCACATCACTAGGGCTCTTACCC
466	C123T	TF	gctggctcggtcaagaGGCCAGGGGCTGGAGGGT
472	A497G	AR	gggacggtcggtagatTCCTCCCGCTGCTTCAGT
472	A497G	EF	GACGATGCCTTCAGCACATCACTTACCCATCATACTCTT TTC
472	A497G	ER	AATCCTGCCTCCACCTT
472	A497G	GR	gctggctcggtcaagaTCCTCCCGCTGCTTCAGC
542	A402G	AR	gggacggtcggtagatAGAAATTCCCTCCCAACT
542	A402G	EF	GACGATGCCTTCAGCACATGATTGAGCCAGTTGTT

baysNP	SNP	Name	Sequence
542	A402G	ER	GGGGTGTATTTGAGAGTG
542	A402G	GR	gctggctcggtcaagaAGAAATTCCCTCCCAACC
739	C87G	CR	gggacggtcggtagatGCTGGTTGACTGGACGG
739	C87G	EF	GACGATGCCTTCAGCACACCTGGTATAATCCTTCC
739	C87G	ER	AGGCAACCTAATCCACTT
739	C87G	GR	gctggctcggtcaagaGCTGGTTGACTGGACGC
821	A140C	AF	gggacggtcggtagatAGTGCTGTGATACTGGA
821	A140C	CF	gctggctcggtcaagaAGTGCTGTGATACTGGC
821	A140C	EF	ACACCCACAAAACAAGAA
821	A140C	ER	GACGATGCCTTCAGCACAGGAACAAGGACATAAAAGAG
1005	A257G	AR	gggacggtcggtagatAGGAAATGTTAGCCCTGT
1005	A257G	EF	GACGATGCCTTCAGCACACTCCACTTCTCTATGCCTC
1005	A257G	ER	GTCCCCAGCTATGTATTGT
1005	A257G	GR	gctggctcggtcaagaAGGAAATGTTAGCCCTGC
1055	A287T	AF	gggacggtcggtagatCTCAGGGAGGGAGAGAGA
1055	A287T	EF	GGGACAGACAGACAGACA
1055	A287T	ER	GACGATGCCTTCAGCACACAACTCCTCTTCAGCAC
1055	A287T	TF	gctggctcggtcaagaCTCAGGGAGGGAGAGAGT
1056	A354G	AR	gggacggtcggtagatGCGGCTGCCCGTCCTGT
1056	A354G	EF	GACGATGCCTTCAGCACAGTGTCTATGTGTCTGTGT
1056	A354G	ER	CGGACTTCTCCTCTTGT
1056	A354G	GR	gctggctcggtcaagaGCGGCTGCCCGTCCTGC
1085	A251G	EF	TAGGGTAAGCAGCAAGAG
1085	A251G	ER	CACAAGGCAAGAGATAACA
1085	A251G	AF	gggacggtcggtagatCAGGCAAGATAGACAGCA
1085	A251G	GF	gctggctcggtcaagaCAGGCAAGATAGACAGCG
1086	A104G	EF	GTGCCCATACGAACAGAATAG
1086	A104G	ER	TGCCAAGTACCCCAAGAG
1086	A104G	AR	gggacggtcggtagatCCATTCCCTCCCCAGACAT
1086	A104G	GR	gctggctcggtcaagaCCATTCCCTCCCCAGACAC
1092	C1687	CF	gggacggtcggtagatCGTGCGAGCAGCGAAAGC

baySNP	SNP	Name	Sequence
	G		
1092	C1687	EF	CCAGAGAGAAGTCGAGGAAGAGA
	G		
1092	C1687	ER	GACGATGCCTTCAGCACAGTCACCCCCAAAAGCAGG
	G		
1092	C1687	GF	gctggctcggtcaagaCGTGCAGCAGCGAAAGG
	G		
1096	G454T	EF	GACGATGCCTTCAGCACACTTTCCCTAGCCCAC
1096	G454T	ER	AAGTGATGTAACCCTCCTCTC
1096	G454T	GR	gggacggtcggtagatTCAGCTATAAAATAGGGCC
1096	G454T	TR	gctggctcggtcaagaTCAGCTATAAAATAGGGCA
1101	C249T	CR	gggacggtcggtagatTGATGGGGGTGCCAAGG
1101	C249T	EF	GACGATGCCTTCAGCACAGCTTTGCTTCC
1101	C249T	ER	CACTGGGGTCCTCTTAC
1101	C249T	TR	gctggctcggtcaagaTGATGGGGGTGCCAAGA
1204	A307G	AR	gggacggtcggtagatCAAGGGCACTCACATTAT
1204	A307G	EF	GACGATGCCTTCAGCACAGCTTGCCTGTTCC
1204	A307G	ER	TTTCCCTCTGCCCCCT
1204	A307G	GR	gctggctcggtcaagaCAAGGGCACTCACATTAC
1504	C180T	CF	gggacggtcggtagatGTGACTTTGGTTCCCAC
1504	C180T	EF	AACTCGGGTCACTGGTCT
1504	C180T	ER	GACGATGCCTTCAGCACACAGCGGGTATGGAGGATG
1504	C180T	TF	gctggctcggtcaagaGTGACTTTGGTTCCCAT
1511	G153T	EF	ACACCAGTTCTCCCTCCT
1511	G153T	ER	GACGATGCCTTCAGCACACCCACCTTCCTAATCCT
1511	G153T	GF	gggacggtcggtagatTTGGGACTCTGCGTCAAG
1511	G153T	TF	gctggctcggtcaagaTTGGGACTCTGCGTCAAT
1524	A284C	AF	gggacggtcggtagatCTCTCAAAGCCCACACAA
1524	A284C	CF	gctggctcggtcaagaCTCTCAAAGCCCACACAC
1524	A284C	EF	AGAAAAAGAAAAGGAAAAAGA
1524	A284C	ER	GACGATGCCTTCAGCACAGGAAAGTTACAAGGCTATGA

baySNP	SNP	Name	Sequence
1556	C367G	CR	gggacggtcggtagatACCTGCCTCTAAGGTCTG
1556	C367G	EF	GACGATGCCTTCAGCACAAAGGAGAACAGTTCAAGG
1556	C367G	ER	ACAGTTGCCAGAGAAAAG
1556	C367G	GR	gctggctcggtcaagaACCTGCCTCTAAGGTCTC
1561	A251C	EF	TCACTTGCCTCTACTCCA
1561	A251C	ER	ATACCAGAAAGACTAAAGCTCC
1561	A251C	AF	gggacggtcggtagatGGGTGAGCTCTGTGGGCA
1561	A251C	CF	gctggctcggtcaagaGGGTGAGCTCTGTGGGCC
1582	C389T	CR	gggacggtcggtagatCCAAGGGTTATGGCAGGG
1582	C389T	EF	GACGATGCCTTCAGCACACCTGACTATTGGGTTGTG
1582	C389T	ER	ATCGCTCTCTGCTTCTGCT
1582	C389T	TR	gctggctcggtcaagaCCAAGGGTTATGGCAGGA
1638	A443G	AR	gggacggtcggtagatCCAAAACCCCAGCGCTGT
1638	A443G	EF	GACGATGCCTTCAGCACACTCTTATCCTGCTTATGGT
1638	A443G	ER	CCAAGCTCACTCTGTAGG
1638	A443G	GR	gctggctcggtcaagaCCAAAACCCCAGCGCTGC
1662	C251T	EF	AATAACAATGGAAGCCAAG
1662	C251T	ER	CCTAACATCGAACAGAAAGG
1662	C251T	CF	gggacggtcggtagatCCAGTCTCCATCCACTTC
1662	C251T	TF	gctggctcggtcaagaCCAGTCTCCATCCACTTT
1714	A376G	AF	gggacggtcggtagatTGAACGGCATGACGGGGA
1714	A376G	EF	AAGTGTTCCTGCTGTGCCT
1714	A376G	ER	GACGATGCCTTCAGCACACAAGTCCTGGTTTCCATC
1714	A376G	GF	gctggctcggtcaagaTGAACGGCATGACGGGG
1722	C89T	CF	gggacggtcggtagatACCCCAGGATGCCACAC
1722	C89T	EF	GTTTATCCTCCTCATGTCC
1722	C89T	ER	GACGATGCCTTCAGCACAGTTACCTTCCACCTCTC
1722	C89T	TF	gctggctcggtcaagaACCCCAGGATGCCACAT
1757	A210G	AF	gggacggtcggtagatGGAAACAAACCAAAATGA
1757	A210G	EF	CCAGCACCCAAAATAAGA
1757	A210G	ER	GACGATGCCTTCAGCACATAAGTTGAAGCCCTCCC

baySNP	SNP	Name	Sequence
1757	A210G	GF	gctggctcggtcaagaGGAAACAAACCAAAATGG
1765	A240G	AF	gggacggtcggtagatGGCTTCACGGAGGAAGAA
1765	A240G	EF	TAGGAGCTGTGAGGTATG
1765	A240G	ER	GACGATGCCTTCAGCACATAAGATGGAGCAGGGTAG
1765	A240G	GF	gctggctcggtcaagaGGCTTCACGGAGGAAGAG
1776	A200G	AF	gggacggtcggtagatAAAGGGCTCCAACACCA
1776	A200G	EF	TGAGCACAAAGATCAGAGAGG
1776	A200G	ER	GACGATGCCTTCAGCACAAAGACAGAGACGCCAGGAATG
1776	A200G	GF	gctggctcggtcaagaAAAGGGCTCCAACACCG
1799	C370T	CF	gggacggtcggtagatAGGGACAACCAAAGTGAC
1799	C370T	EF	ATCATCAGAACAGCCCTAC
1799	C370T	ER	GACGATGCCTTCAGCACACAAGCCCACCTACTTACTC
1799	C370T	TF	gctggctcggtcaagaAGGGACAACCAAAGTGAT
1806	A201G	AF	gggacggtcggtagatTGGCGTCCCTGGTGGGCA
1806	A201G	EF	TCTTCGGGCTAACTCTTT
1806	A201G	ER	GACGATGCCTTCAGCACACTGTCACTCCAAACCTTCT
1806	A201G	GF	gctggctcggtcaagaTGGCGTCCCTGGTGGCG
1837	C413T	CF	gggacggtcggtagatCTCAGCTTCATGCAGGGC
1837	C413T	EF	CCCACTCAGCCCTGCTCTT
1837	C413T	ER	GACGATGCCTTCAGCACAGCATCCTGGCGGTCTTG
1837	C413T	TF	gctggctcggtcaagaCTCAGCTTCATGCAGGGT
1870	C323T	CF	gggacggtcggtagatCTCCTCATTGCCTCCTTC
1870	C323T	EF	CACCTCTTTCTCCTTCTCTT
1870	C323T	ER	GACGATGCCTTCAGCACACCCACCCCTCTATCTAC
1870	C323T	TF	gctggctcggtcaagaCTCCTCATTGCCTCCTTT
1882	C115T	CR	gggacggtcggtagatGTCCCCACAGTCCTCG
1882	C115T	EF	GACGATGCCTTCAGCACAGACCTGTACCCCTTACCC
1882	C115T	ER	TGTTCCCTGTCTGTTTC
1882	C115T	TR	gctggctcggtcaagaGTCCCCACAGTCCTCA
1988	C214T	CF	gggacggtcggtagatGTGACTCGGTCCATTACCC
1988	C214T	EF	GTGGGCTGTGATTGTGTT

baySNP	SNP	Name	Sequence
1988	C214T	ER	GACGATGCCTTCAGCACATCTCGTCGTAGTAGTTGT
1988	C214T	TF	gctggctcggtcaagaGTGACTCGGTCCCTATACT
2000	C349T	CR	gggacggtcggtagatAGTATGGTAATTAGGAAG
2000	C349T	EF	GACGATGCCTTCAGCACACTGACACTGAGCCACAAC
2000	C349T	ER	AACTGATGAGCAAGAAGGA
2000	C349T	TR	gctggctcggtcaagaAGTATGGTAATTAGGAAA
2071	A338G	AR	gggacggtcggtagatAAAATTGTTCCCTGTGAT
2071	A338G	EF	GACGATGCCTTCAGCACACATTGCTATTCTCAGGCTATA
2071	A338G	ER	CCCATTCTCTGCTTGACAGT
2071	A338G	GR	gctggctcggtcaagaAAAATTGTTCCCTGTGAC
2078	G876T	EF	CCAGAGAGGGATAAAAGA
2078	G876T	ER	GACGATGCCTTCAGCACAGAGTGTCAAGAGGAACAGG
2078	G876T	GF	gggacggtcggtagatTGGCTGCTGAGGTCTGAG
2078	G876T	TF	gctggctcggtcaagaTGGCTGCTGAGGTCTGAT
2085	G415T	EF	GCTTTTCTTTCATACATC
2085	G415T	ER	GACGATGCCTTCAGCACACCTTTAGAACAGAGACA
2085	G415T	GF	gggacggtcggtagatGGTAGTGTACCAGAAAG
2085	G415T	TF	gctggctcggtcaagaGGTAGTGTACCAGAAAT
2095	A406G	AR	gggacggtcggtagatTGTGCACCGGGATATTT
2095	A406G	EF	GACGATGCCTTCAGCACAAATGTGTGCTGGTTCTT
2095	A406G	ER	GGTGTTCCTCCCTCTCT
2095	A406G	GR	gctggctcggtcaagaTGTGCACCGGGATATTTC
2119	A67G	AR	gggacggtcggtagatGTGGGCACCAAACGCTAT
2119	A67G	EF	GACGATGCCTTCAGCACAGATGTAGGGCTGGAAGTG
2119	A67G	ER	TCAAGAAAATGGGAGTTG
2119	A67G	GR	gctggctcggtcaagaGTGGGCACCAAACGCTAC
2141	A176G	EF	TGTAGCATCGGTAGGTTC
2141	A176G	ER	CAACATCAGACTTTCTTTTC
2141	A176G	AR	gggacggtcggtagatTGGTACAGGGCTAGTTTT
2141	A176G	GR	gctggctcggtcaagaTGGTACAGGGCTAGTTTC
2182	A318G	AF	gggacggtcggtagatAGGCGGGCCAAGGGTGAA

baySNP	SNP	Name	Sequence
2182	A318G	EF	TTCTCTCCTCCCTTCTGT
2182	A318G	ER	GACGATGCCTTCAGCACATAAATGTTCACTCTTCTTGCT
2182	A318G	GF	gctggctcggtcaagaAGGCGGGCCAAGGGTGAG
2234	G296T	EF	GGGTTGTTCCAGGGCGCTATT
2234	G296T	ER	GACGATGCCTTCAGCACATGTGGAGAGGCCGGTGC
2234	G296T	GF	gggacggtcggtagatGAACCAGCCCCCTGGAAG
2234	G296T	TF	gctggctcggtcaagaGAACCAGCCCCCTGGAAT
2281	A227C	AR	gggacggtcggtagatCAGGCTTGGAGACCTGGT
2281	A227C	CR	gctggctcggtcaagaCAGGCTTGGAGACCTGGG
2281	A227C	EF	GACGATGCCTTCAGCACAGGGTATTCAAGTTGGAAGG
2281	A227C	ER	AAGGCAAGGTTCTTAGTTG
2298	A77C	AR	gggacggtcggtagatTCTAAAAGCACTTGAAAT
2298	A77C	CR	gctggctcggtcaagaTCTAAAAGCACTTGAAAG
2298	A77C	EF	GACGATGCCTTCAGCACACCTGCTAGTGTGTTCTGG
2298	A77C	ER	TGTAAC TGATAGGTGGTGG
2341	C286T	CR	gggacggtcggtagatTGAAGATTCTGCTCAGCG
2341	C286T	EF	GACGATGCCTTCAGCACAAAGGCCGGACTCAT
2341	C286T	ER	TTTGGGGTCCTGCGGATG
2341	C286T	TR	gctggctcggtcaagaTGAAGATTCTGCTCAGCA
2357	A165G	AF	gggacggtcggtagatCAAAGAACGAAATGA
2357	A165G	EF	CTCAAGTTGTTACTGATTCTC
2357	A165G	ER	GACGATGCCTTCAGCACAGGGTACGTCTGCTCTTC
2357	A165G	GF	gctggctcggtcaagaCAAAGAACGAAATGG
2366	G50T	EF	GACGATGCCTTCAGCACACTGCTCCGAAACACGGTC
2366	G50T	ER	GCATCTTCAGCCCTTCTTACTCT
2366	G50T	GR	gggacggtcggtagatCTCCTGGGCACCACGGGC
2366	G50T	TR	gctggctcggtcaagaCTCCTGGGCACCACGGGA
2995	A299C	ER	gacgatgccttcagcacaTGGGATTAGACACGAGAG
2995	A299C	EF	AAAGAACTGGAAGAACGAA
2995	A299C	AF	gggacggtcggtagatGTCACCTCCTTCCACTA
2995	A299C	CF	gctggctcggtcaagaGTCACCTCCTTCCACTC

baySNP	SNP	Name	Sequence
3360	G777T	ER	gacgatgccttcagcacaAGAAAAATGAGAGGGAAAAC
3360	G777T	EF	GATGAAGGGAAATGGAAC
3360	G777T	GF	gggacggtcggttagatCCAACCTATATAGGAGCCG
3360	G777T	TF	gctggctcggtcaagaCCAACCTATATAGGAGCCT
3464	A110G	EF	CTGAACCGAGGAGATTTT
3464	A110G	ER	TGATGCTTACAGAACTGGG
3464	A110G	AF	gggacggtcggttagatGTGTAGTGGGCAGGGTTA
3464	A110G	GF	gctggctcggtcaagaGTGTAGTGGGCAGGGTTG
3975	A65C	EF	gacgatgccttcagcacaAAAAGAACCTGGTGAAG
3975	A65C	ER	CCCTGATAAAAGAGATGGA
3975	A65C	AR	gggacggtcggttagatCGCATGGGAGTCAGGGAT
3975	A65C	CR	gctggctcggtcaagaCGCATGGGAGTCAGGGAG
3976	A239G	EF	gacgatgccttcagcacaATGAGGGAGCAAGACAAG
3976	A239G	ER	TGATAAAAGAGATGGAAGGGAG
3976	A239G	AR	gggacggtcggttagatGTCACTGTTGTCACTGT
3976	A239G	GR	gctggctcggtcaagaGTCACTGTTGTCACTGC
4206	A304T	EF	gacgatgccttcagcacaCTTTTAGCCAAGTGGAG
4206	A304T	ER	GGATCTGAGGAATCTGTG
4206	A304T	AR	gggacggtcggttagatACCAGGCAGAGAGAAAAAT
4206	A304T	TR	gctggctcggtcaagaACCAGGCAGAGAGAAAAAA
4912	A74G	EF	CTTCACTGAGCGTCCGCAGAG
4912	A74G	ER	CCGTCGGCCCGATTCA
4912	A74G	AR	CAGGCGAGCCTCAGCCCT
4912	A74G	GR	CAGGCGAGCCTCAGCCCC
4925	A251C	EF	TCATTTCCAATTACCTCC
4925	A251C	ER	CCTCTTCCCATCTCCCT
4925	A251C	AF	gggacggtcggttagatAGCCAGGAGCCTGCGTCA
4925	A251C	CF	gctggctcggtcaagaAGCCAGGAGCCTGCGTCC
4966	A251G	EF	CATTGCTCTCCTCTGT
4966	A251G	ER	GTGTCATCATTCCCTTCTTG
4966	A251G	AR	gggacggtcggttagatTCAGAGACATGAGTCCAT

baySNP	SNP	Name	Sequence
4966	A251G	GR	gctggctcggtcaagaTCAGAGACATGAGTCCAC
5014	A2057	ER	gacgatgccttcagcacaCACCTGTCCCACCCCTATTG
5014	A2057	EF	GTCCTGAACCCCCATTCT
5014	A2057	AF	gggacggtcggtagatGCCTGCACTGCGTTCTA
5014	A2057	GF	gctggctcggtcaagaGCCTGCACTGCGTTCTG
5296	A251G	EF	GCTCCTCTGCCTTCTGCTT
5296	A251G	ER	ATTTGCCCACTGCCCTTC
5296	A251G	AF	gggacggtcggtagatTGGCTGCAGGTGCGTCCA
5296	A251G	GF	gctggctcggtcaagaTGGCTGCAGGTGCGTCCG
5298	C172T	EF	GCCACACACACCTTAACA
5298	C172T	ER	AAAGTTCTCTGCCTCCAA
5298	C172T	CF	gggacggtcggtagatAGCTCTCAGCTGGGGTGC
5298	C172T	TF	gctggctcggtcaagaAGCTCTCAGCTGGGGTGT
5457	A134G	EF	AGCAGAACGGCAATAGA
5457	A134G	ER	AGAGATGTGGCAGAGAA
5457	A134G	AF	gggacggtcggtagatGGAAAGCCTACTTTCTTA
5457	A134G	GF	gctggctcggtcaagaGGAAAGCCTACTTTCTTG
5704	C61T	EF	ACAGCCATAACAGGAGTG
5704	C61T	ER	GGGTTACTCAACCTAACAGAGA
5704	C61T	CR	gggacggtcggtagatGTTCTCTTGGAAAACG
5704	C61T	TR	gctggctcggtcaagaGTTCTCTTGGAAAACA
5717	A1960	EF	gacgatgccttcagcacaAACAGAAACCACAGAACCG
5717	A1960	ER	GTCCCACCCCTATTTGAG
5717	A1960	AR	gggacggtcggtagatCACTGGCCCACCTCCCTT
5717	A1960	GR	gctggctcggtcaagaCACTGGCCCACCTCCCTC

baySNP	SNP	Name	Sequence
	G		
5959	A71G	EF	gacgatgccttcagcacaACCATGCCTGACTTAACC
5959	A71G	ER	TTGTTTCCTGTCCTCTTTC
5959	A71G	AR	gggacggtcggtagatGTTAAGAGGCTGGCAGT
5959	A71G	GR	gctggctcggtcaagaGTTAAGAGGCTGGCAGC
6162	C340G	EF	gacgatgccttcagcacaAGTGTGTTAGGAGCAAAG
6162	C340G	ER	CTTAGGAAACTGAGGTGG
6162	C340G	CR	gggacggtcggtagatCTGCAGCCTGGCAACAG
6162	C340G	GR	gctggctcggtcaagaCTGCAGCCTGGCAACAC
6236	C906T	ER	gacgatgccttcagcacaTGGACACATTGAGCTT
6236	C906T	EF	CTTCCCCAGAGATGACTAC
6236	C906T	CF	gggacggtcggtagatCCCCATCCTACTCAGCAC
6236	C906T	TF	gctggctcggtcaagaCCCCATCCTACTCAGCAT
6744	C348T	ER	gacgatgccttcagcacaGGTTACAGTGAGCCAAGA
6744	C348T	EF	AGGTGAAGAAAGCAAAATAC
6744	C348T	CF	gggacggtcggtagatGGGTGTGTGTTTGTTC
6744	C348T	TF	gctggctcggtcaagaGGGTGTGTGTTTGTTT
7133	C63G	EF	TTGAGACCCTACAGAGCCA
7133	C63G	ER	GGCAAGCTGAGGTGAAAG
7133	C63G	CR	gggacggtcggtagatATAAGGTAAGAAATGAG
7133	C63G	GR	gctggctcggtcaagaATAAGGTAAGAAATGAC
8210	A251G	EF	TAATTCTAATGGCCTTCC
8210	A251G	ER	TCACTTACTCCCTGATGTCT
8210	A251G	AR	gggacggtcggtagatCATTGGGTTTCCTCAT
8210	A251G	GR	gctggctcggtcaagaCATTGGGTTTCCTCAC
8592	C46T	ER	gacgatgccttcagcacaACATTAGTGCCAACATCAC
8592	C46T	EF	CTCTTCCTGAGACACCA
8592	C46T	CF	gggacggtcggtagatGAAGGTGAAGGCCAGAGC
8592	C46T	TF	gctggctcggtcaagaGAAGGTGAAGGCCAGAGT
8943	A251C	EF	GAGGCTGAGACAGAAGAA
8943	A251C	ER	GTTCGACATTAAAGAAAATGAG

baySNP	SNP	Name	Sequence
8943	A251C	AR	gggacggtcggtagatGGCTGGAGTGCAGTGATT
8943	A251C	CR	gctggctcggtcaagaGGCTGGAGTGCAGTGATG
9193	C88G	EF	CACGCTGTTGAGTGGG
9193	C88G	ER	CGCAGGTCTACGGTCA
9193	C88G	CR	gggacggtcggtagatCCCGGGTCTGAGGCTGCG
9193	C88G	GR	gctggctcggtcaagaCCCGGGTCTGAGGCTGCC
9516	A187G	EF	CACACACACACACACACAC
9516	A187G	ER	GGTCCCTTACTTCCTCTT
9516	A187G	AR	gggacggtcggtagatCCTATCCCTACTTCCCC
9516	A187G	GR	gctggctcggtcaagaCCTATCCCTACTTCCCC
9698	A251G	EF	GTGACCCCCAAAAGAGAGA
9698	A251G	ER	CTAGCTTGTACTGCCTCC
9698	A251G	AF	gggacggtcggtagatGGCACGACCCGCCCG
9698	A251G	GF	gctggctcggtcaagaGGCACGACCCGCCCG
9883	A249G	EF	TCCACAAACCTCAAACCAC
9883	A249G	ER	CACAGTCCTGCAAGCTCA
9883	A249G	AR	gggacggtcggtagatCCGTGGCCGTGGCTCACT
9883	A249G	GR	gctggctcggtcaagaCCGTGGCCGTGGCTCACC
10481	A107T	ER	gacgatgccttcagcacAGTCGGGGCTCCACTT
10481	A107T	EF	TAGCGGGACAGCGCTG
10481	A107T	AF	gggacggtcggtagatCCCGGCGCGCCTCGGAGA
10481	A107T	TF	gctggctcggtcaagaCCCGGCGCGCCTCGGAGT
10542	C367T	EF	gacgatgccttcagcacAAATACACTGGGTCTGCT
10542	C367T	ER	ATACTGCTGGCCTTCCTC
10542	C367T	CR	gggacggtcggtagatGGTCAGGGGAGCCCAGAG
10542	C367T	TR	gctggctcggtcaagaGGTCAGGGGAGCCCAGAA
10600	A251G	EF	CCTGGCAACTAACCTCTT
10600	A251G	ER	AGGCAGTCTCTGTCTACTC
10600	A251G	AR	gggacggtcggtagatATTGGCCCTGCTCAGGAT
10600	A251G	GR	gctggctcggtcaagaATTGGCCCTGCTCAGGAC
10621	C402T	EF	CCAGCCCTAAACCTAAA

baySNP	SNP	Name	Sequence
10621	C402T	ER	AACCTCTCAAGATCAGACAC
10621	C402T	CF	gggacggtcggtagatTTAGCACTTAATAAGTAC
10621	C402T	TF	gctggctcggtcaagattAGCACTTAATAAGTAT
10745	A251G	EF	CCCCACAACAAAGAAAGA
10745	A251G	ER	GAAGCCAACCTCTCCAACA
10745	A251G	AF	gggacggtcggtagatCAAGGATTCAAAAACCA
10745	A251G	GF	gctggctcggtcaagaCAAGGATTCAAAAACCG
10771	C64G	EF	gacgatgccttcagcacaCCAGGGAAAGAGCAGAAC
10771	C64G	ER	TGTACGGGAAGAGGCAGA
10771	C64G	CR	gggacggtcggtagatAGGGTGACACAGGCCACG
10771	C64G	GR	gctggctcggtcaagaAGGGTGACACAGGCCACC
10870	A251G	EF	ATCCCATCCCAACACACA
10870	A251G	ER	CCGAGACCAAACTCATTAC
10870	A251G	AR	gggacggtcggtagatGGCAGAGCCTGAGTCACT
10870	A251G	GR	gctggctcggtcaagaGGCAGAGCCTGAGTCACC
10877	A251C	EF	CCTGTTCTCAACCTTCTC
10877	A251C	ER	ATGGTCTATGGAACCTAATCT
10877	A251C	AF	gggacggtcggtagatGCACTGATTCTGCTTCCA
10877	A251C	CF	gctggctcggtcaagaGCACTGATTCTGCTTCCC
10948	G140T	EF	AAGGACAGGGTCAGGAAAG
10948	G140T	ER	CAGAGGGAGGAAGGAGGT
10948	G140T	GF	gggacggtcggtagatATGGAGGAGGGTGTCTGG
10948	G140T	TF	gctggctcggtcaagaATGGAGGAGGGTGTCTGT
11001	C286T	EF	gacgatgccttcagcacaTTCCCAAAGACCCACA
11001	C286T	ER	CCTCCACCGCTATCAC
11001	C286T	CR	gggacggtcggtagatTGGCTGCAGGACGTCCAG
11001	C286T	TR	gctggctcggtcaagaTGGCTGCAGGACGTCCAA
11001	C286T	EF	TTCCCAAAGACCCACA
11001	C286T	ER	CCTCCACCGCTATCAC
11001	C286T	CR	gggacggtcggtagatTGGCTGCAGGACGTCCAG
11001	C286T	TR	gctggctcggtcaagaTGGCTGCAGGACGTCCAA

baySNP	SNP	Name	S quence
11073	C215G	EF	CCCAACCACCCGGTCC
11073	C215G	ER	GCGCGGGAGCTAGAGA
11073	C215G	CF	gggacggtcggtagatGAAGCTGCAGGCCGGAC
11073	C215G	GF	gctggctcggtcaagaGAAGCTGCAGGCCGGACG
11153	C116T	EF	CGAGTGGGAAGAAAAGTAGA
11153	C116T	ER	ATGACTGCCTGCCTAGAA
11153	C116T	CR	gggacggtcggtagatAAGATAGGGTAGAGGCCG
11153	C116T	TR	gctggctcggtcaagaAAGATAGGGTAGAGGCCA
11210	C194T	EF	GAGGAGTGAGGGAAAGTAAG
11210	C194T	ER	AAATGGAGAGAGATGGGA
11210	C194T	CF	gggacggtcggtagatCCAGGAAATGACATGATC
11210	C194T	TF	gctggctcggtcaagaCCAGGAAATGACATGATT
11248	C225T	EF	TGAGTTAACAGCACTTGG
11248	C225T	ER	AGGGTAAGGGAGGGAAAA
11248	C225T	CR	gggacggtcggtagatTGATTCTTCGCTTGGCG
11248	C225T	TR	gctggctcggtcaagaTGATTCTTCGCTTGGCA
11372	A251G	EF	TAGAAAAGAAGAAAAATCAA
11372	A251G	ER	ACACACACACACACACAC
11372	A251G	AR	gggacggtcggtagatCATCACCTTTAGTTCT
11372	A251G	GR	gctggctcggtcaagaCATCACCTTTAGTTCC
11449	C251G	EF	ACAGAAGAACAAACAACAAAC
11449	C251G	ER	TGCGTATGAGGTAAAGAGA
11449	C251G	CF	gggacggtcggtagatATGAGTGAAGCCTGTCTC
11449	C251G	GF	gctggctcggtcaagaATGAGTGAAGCCTGTCTG
11450	A251T	EF	ACAGAAGAACAAACAACAAAC
11450	A251T	ER	TGCGTATGAGGTAAAGAGA
11450	A251T	AR	gggacggtcggtagatGGACCATAATCTGAAGT
11450	A251T	TR	gctggctcggtcaagaGGACCATAATCTGAAGA
11470	C251T	EF	GCTTGTCTGTCTGATAGGTG
11470	C251T	ER	CAACGTGAGAATTCCAAAAT
11470	C251T	CR	gggacggtcggtagatTGAGAATTCCAAAATAG

baySNP	SNP	Name	Sequence
11470	C251T	TR	gctggctcggtcaagaTGAGAATTCCAAAATAA
11472	A251T	EF	TACATTCAAGGCAAGAAAA
11472	A251T	ER	TGATTAGTTACAATTACCTCTAGTATC
11472	A251T	AF	gggacggtcggtagatAGTTGTCACTAAATGTA
11472	A251T	TF	gctggctcggtcaagaAGTTGTCACTAAATGTT
11487	A485T	EF	gacgatgccttcagcacaAGAGAGCAGCTAGACTGAGA
11487	A485T	ER	TTCCTGCAAACAGTTGAG
11487	A485T	AR	gggacggtcggtagatAGTTGAGGGCTCAGGATT
11487	A485T	TR	gctggctcggtcaagaAGTTGAGGGCTCAGGATA
11488	C533G	EF	gacgatgccttcagcacaAGAGAGCAGCTAGACTGAGA
11488	C533G	ER	GTAAATAAAATGGGATGGT
11488	C533G	CR	gggacggtcggtagatGCCAGCAAGCTGCATG
11488	C533G	GR	gctggctcggtcaagaGCCAGCAAGCTGCATC
11493	A171G	EF	CCTTTGTGTTTGT
11493	A171G	ER	CTTCTCCACCTTCCATTC
11493	A171G	AF	gggacggtcggtagatGGGAACCTCTAAATCAAA
11493	A171G	GF	gctggctcggtcaagaGGGAACCTCTAAATCAAG
11502	C455T	EF	gacgatgccttcagcacaACGATGGGTCAGAGTCA
11502	C455T	ER	CCTACATTCACACACGAACA
11502	C455T	CR	gggacggtcggtagatACACACTCCTCTCAAG
11502	C455T	TR	gctggctcggtcaagaACACACTCCTCTCAAA
11534	G258T	EF	GCCATCGTCTTCCCT
11534	G258T	ER	TCCTCCCTCCTCTCTCT
11534	G258T	GR	gggacggtcggtagatCCTCCACCCACCAGGGCC
11534	G258T	TR	gctggctcggtcaagaCCTCCACCCACCAGGGCA
11537	A251G	EF	CCTTTCTCCTCTTC
11537	A251G	ER	CTCTTCCCTGTCTTCTCCTCT
11537	A251G	AF	gggacggtcggtagatAGATGGACCTCTACAGGA
11537	A251G	GF	gctggctcggtcaagaAGATGGACCTCTACAGGG
11560	A185G	EF	CTCCTCCAACTCCTTAC
11560	A185G	ER	ATACTTCTCACTGCATCCT

baySNP	SNP	Name	Sequence
11560	A185G	AR	gggacggtcggtagatCCTGTCCCCTCCCTAGTT
11560	A185G	GR	gctggctcggtcaagaCCTGTCCCCTCCCTAGTC
11594	C251T	EF	CACCTTCCTGAACTCACTC
11594	C251T	ER	TGATGTCTGTGCTGTCT
11594	C251T	CR	gggacggtcggtagatTCTGGTCCACTCAAGGAG
11594	C251T	TR	gctggctcggtcaagaTCTGGTCCACTCAAGGAA
11624	C251T	EF	TCGGGAGGTGTAAGTAAG
11624	C251T	ER	CCACAGTCAGAAGAGACAA
11624	C251T	CR	gggacggtcggtagatAGAGACCCTGGTCCAAG
11624	C251T	TR	gctggctcggtcaagaAGAGACCCTGGTCCAAA
11627	C251T	EF	TTTATCACTACACCCCCCTACTC
11627	C251T	ER	GACAGACCGACCAATCAC
11627	C251T	CR	gggacggtcggtagatCCCTGGGAAGGTTGAGAG
11627	C251T	TR	gctggctcggtcaagaCCCTGGGAAGGTTGAGAA
11650	A146G	EF	CTGTCTGTTGGTCTTC
11650	A146G	ER	CGTTGTTCTCTGTCCACT
11650	A146G	AR	gggacggtcggtagatGCCAAATGTCTAAAAGT
11650	A146G	GR	gctggctcggtcaagaGCCAAATGTCTAAAAGC
11654	A251G	EF	CGTATCTTGCCTTCTT
11654	A251G	ER	CTTCTTTATGCCTTCCC
11654	A251G	AF	gggacggtcggtagatTTACTTGAAAGGACACCA
11654	A251G	GF	gctggctcggtcaagaTTACTTGAAAGGACACCG
11655	A251C	EF	CGTATCTTGCCTTCTT
11655	A251C	ER	CTTCTTTATGCCTTCCC
11655	A251C	AF	gggacggtcggtagatTTCTGCACTAAAGCTGTA
11655	A251C	CF	gctggctcggtcaagaTTCTGCACTAAAGCTGTC
11656	C251T	EF	TGGGAAGAAAAAGAGAAG
11656	C251T	ER	GTTGAAACACTGCACAAG
11656	C251T	CR	gggacggtcggtagatCAGGGCTGTTGGGTGAAG
11656	C251T	TR	gctggctcggtcaagaCAGGGCTGTTGGGTGAAA
11825	A277G	ER	gacgatgcctcagcacatGAATAGACAGGGACGAA

baySNP	SNP	Name	Sequence
11825	A277G	EF	GACCTTGGAAATAATGGAG
11825	A277G	AF	gggacggtcggtagatCAACCCAGCAAAAATGGA
11825	A277G	GF	gctggctcggtcaagaCAACCCAGCAAAAATGGG
11914	A246T	EF	gacgatgccttcagcacaTTGGAAGTGAGATAAGATAGGT
11914	A246T	ER	ACGGTGAGAATGAGAGGT
11914	A246T	AR	gggacggtcggtagatAAAACAGACATCAGAGGT
11914	A246T	TR	gctggctcggtcaagaAAAACAGACATCAGAGGA
12097	A411G	ER	gacgatgccttcagcacaGATGAAACCCTGTCTACT
12097	A411G	EF	TTATCACACCTTAGTCTCCCT
12097	A411G	AF	gggacggtcggtagatACCTGCCACACACCAA
12097	A411G	GF	gctggctcggtcaagaACCTGCCACACACCCAG
12366	A412G	ER	gacgatgccttcagcacaGCTGATGTGGTTGTGAG
12366	A412G	EF	GTTCCCTGTAGCTCGTGTAG
12366	A412G	AF	gggacggtcggtagatCTCCCCGCCCTGCAGCAA
12366	A412G	GF	gctggctcggtcaagaCTCCCCGCCCTGCAGCAG
12619	A25G	ER	gacgatgccttcagcacaTGGCTGGACTTGACTGATA
12619	A25G	EF	TCTTGTGTCACAGTGC
12619	A25G	AF	gggacggtcggtagatTGTGTCACAGTGCTCTGA
12619	A25G	GF	gctggctcggtcaagaTGTGTCACAGTGCTCTGG
13025	A585C	EF	gacgatgccttcagcacaTTAACGTAACATGACAAACTC
13025	A585C	ER	ATCTGATAACTGAGCAGG
13025	A585C	AR	gggacggtcggtagatCTATTAAAGTAACACTGGTGT
13025	A585C	CR	gctggctcggtcaagaCTATTAAAGTAACACTGGTGG
13191	A504G	ER	gacgatgccttcagcacaATTCTCCATTCTCCTGT
13191	A504G	EF	TGCCTCTTCTCCTCATTC
13191	A504G	AF	gggacggtcggtagatCCCTAATGTCTTCCTCTGA
13191	A504G	GF	gctggctcggtcaagaCCCTAATGTCTTCCTCTGG
900045	C116T	EF	ATCTCCTGATCCAAGTCC
900045	C116T	ER	CACACTGTGCCCATCTAC
900045	C116T	CR	gggacggtcggtagatCTGACTGATTACCTCATG
900045	C116T	TR	gctggctcggtcaagaCTGACTGATTACCTCATA

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900078	A251G	EF	CATAGGTAAAGATCTGTAGGTG
900078	A251G	ER	CCACCTTGGAAAGTTGGCAAA
900078	A251G	AR	gggacggtcggtagatattaaatgcctctctcT
900078	A251G	GR	gctggctcggtcaagaattaaatgcctctctcC
900107	C426T	ER	gacgatgccttcagcacaAGGGCTTTTCAGGTAGA
900107	C426T	EF	GACCTTCCTGGGTAGAA
900107	C426T	CF	gggacggtcggtagatACTCTGAACCTGGGGAC
900107	C426T	TF	gctggctcggtcaagaACTCTGAACCTGGGGAT
10000002	A103G	AF	gggacggtcggtagatGATCAACACAATCTCAA
10000002	A103G	EF	CAGCTGAAAGAGATGAAATTACT
10000002	A103G	ER	GACGATGCCTTCAGCACAAACTTATGAAGATTAAAGGCATAG G
10000002	A103G	GF	gctggctcggtcaagaGATCAACACAATCTCAG
10000006	G107A	AF	gctggctcggtcaagaGGGCTGGGCTGCTAGGGAA
10000006	G107A	EF	AGACGAGTTCAAGGTGAGTG
10000006	G107A	ER	GACGATGCCTTCAGCACACCAAGTTCCGAGTTCC
10000006	G107A	GF	gggacggtcggtagatGGGCTGGGCTGCTAGGGG
10000014	A153C	AF	gggacggtcggtagatGTACCAATACATCCTGCA
10000014	A153C	CF	gctggctcggtcaagaGTACCAATACATCCTGCC
10000014	A153C	EF	CTGCTGATGTCTCTGTTG
10000014	A153C	ER	GACGATGCCTTCAGCACAGACTTACTTGCTCACACTT
10000025	C291T	CF	gggacggtcggtagatCCTCACTCCTAACGCC
10000025	C291T	EF	CCTCTCTGCTGGTTATCTTG
10000025	C291T	ER	GACGATGCCTTCAGCACAAAGTGTGCCTCCTGGTTAG
10000025	C291T	TF	gctggctcggtcaagaCCTCACTCCTAACGCT

Table 2b Oligonucleotide primers used for genotyping using Pyrosequencing

The baySNP number refers to an internal numbering of the PA SNPs. Primer sequences are listed for preamplification of the genomic fragments and for sequencing of the SNP using the pyrosequencing method. Bio: Biotinylated Oligonucleotide.

BaySNP	Name	Sequence
2995	Primer F	GCCAAGACTAGGAAGTAAGTGT
2995	Primer R	Bio- CCCAGAACACAAAGCTAGTAA
2995	Seq.	TGCCCTGGTCACCTCCTTCC
3689	Primer F	BIO-CTGACCCTGACCTTCATACTCAA
3689	Primer R	AGAAGAAAGAACGCCTCTACAGTT
3689	Seq.	AACAGATCAGGTTGGTG
4838	Primer F	Bio-CAAAGATGACCTTATGGCTCTGA
4838	Primer R	GTCTCGGAACATGACCTTTAGT
4838	Seq.	TGACTAAGAATGTAATGGGGAAGA
6498	Primer F	CTTGTGGATCTTCTGCGGTGT
6498	Primer R	Bio-CCATGTTGAGGAGCCCAGAGTGA
6498	Seq.	ATTACAGTTGTGAGATTGTGC
8021	Primer F	GGCCTCTATGTACTAGGCG
8021	Primer R	Bio-CTCTTCTGGAGGCATCAATC
8021	Seq.	CACAGGGAGACCCC
8060	Primer F	Bio-GCCTTATTTCCACTCCACCT
8060	Primer R	TACCTTCCCCATCCAACTG
8060	Seq.	TCAGCATATGTTGGATT
8846	Primer F	ATTTGAGAGAAGGTAGGGT
8846	Primer R	BIO-TTTGTTACTCTGTAGCCA
8846	Seq.	AAATATTCACTTGT
9849	Primer F	AAG CAG CAA TCG AAT CCC TT
9849	Primer R	TGT TGT TGT TTG GCT AGC TCC
9849	Seq.	CCT GCC TTA CTG AGA GCC AAA

BaySNP	Name	Sequence
10079	Primer F	Bio-CACGCCAATTCCCCACCATCCT
10079	Primer R	GTCCGTCGAGGGGGAAATGTGTTT
10079	Seq.	AATGTGTTCTTGGGGGT
10747	Primer F	CTAACCATCTTCAAATGCTTAATC
10747	Primer R	BIO-TCCTTGAGTCTGAGTTCCC
10747	Seq.	CACAAGAAACCCTGAAA
11578	Primer F	CTC GGC GTG CTT GGT AAT AA
11578	Primer R	CGG AGC CGA ACT CTG GAG GAA TCT
11578	Seq.	GGC TGG CAA GTT GTT CCA TCC CAC
11644	Primer F	TGA GCA GCG CAT CCT
11644	Primer R	TGC AGC CCA CTG ACT CAA
11644	Seq.	GCT GTT ACT CAG TAT GAT
12008	Primer F	CCGAAGACCAAGACGC
12008	Primer R	Bio-TCTTCCATAAAAACAAGGCTC
12008	Seq.	AAACAAGAAATTCTGTTA
13937	Primer F	TGA CAG CTC CCA TTG GAA
13937	Primer R	AAT TAA TGC GAT CCC TC
13937	Seq.	GAC AGC TCC CAT TGG AAG
900002	Primer F	ATTGGGCAGGGATAAGAGAAAAG
900002	Primer R	Bio-GATGAATCACAGAATGCGGTAT
900002	Seq.	CACACAGCAGTTCACGCA
900013	Primer F	GCCAAGACTAGGAAGTAAGTGT
900013	Primer R	Bio- CCCAGAACACAAAGCTAGTAA
900013	Seq.	TGCCCTGGTCACCTCCTTCC
900025	Primer F	Bio-AGTGGCTCACCTGCTAACG
900025	Primer R	CTGGGGAAGAAAATAATGAA
900025	Seq.	CTTGCTCTTAGGATACACGT
900032	Primer F	AGCGTCTCACCATCTGCT
900032	Primer R	Bio-GGGAAGGAGGAAGCCAAACA
900032	Seq.	ACATGTCTGATGATACCTGG
900045	Primer F	BIO-GCCATGCACGATTCGG

BaySNP	Name	Sequence
900045	Primer R	CACTGTGCCCATCTACGAG
900045	Seq.	GGACCTGACTGATTACCT
900065	Primer F	GAGTAGCTAGGATCACAGGTGCGT
900065	Primer R	BIO-TGTCGAGATTAAAGAAAGTTGGC
900065	Seq.	CAGGTGCGTGCCACCATGCC
900082	Primer F	CAC ACA ATT TTC CAC TTA
900082	Primer R	GAC TCC AGT TTT CTA TCA
900082	Seq.	ATG TTG ATG TAA TCT ACT
900096	Primer F	TGGGGCAAGAACAGTGGT
900096	Primer R	Bio-TAGGCAGGGCAAGGGATTAGG
900096	Seq.	TTTAAATTCTCTGACAGAGAC
900107	Primer F	BIO-GCCACCAGCCCACACTCTGAACCTG
900107	Primer R	CCATCAGCCTTCACCCACGTGCCA
900107	Seq.	GCCTCAGCTTGACCT
900115	Primer F	Bio-GGTAAGTGCCTGCCTGGGAGATGC
900115	Primer R	CGGGGTGGGGAGGGACAGAGC
900115	Seq.	GAGGACAGAGCAAAAGGAT
900121	Primer F	Bio-TGCCTTACAATATAACATGG
900121	Primer R	CAATGGGTAAGGAGTAAAGTT
900121	Seq.	TTCCAGCTGCTTTA

Table 2c Oligonucleotide primers used for genotyping using Restriction Fragment Length Polymorphism (RFLP)

- 5 The baySNP number refers to an internal numbering of the PA SNPs. Primer sequences are listed for preamplification of the genomic fragments. The restriction enzyme used for RFLP is indicated.

baySNP	Name	Sequence	Enzyme
900173	Primer F	GAACAAACCTCCGAGATGCTAC	Hind III
900173	Primer R	GTCTTATGTTACTGGGCTTCACC	Hind III

Table 2d Oligonucleotide primers used for genotyping using TaqMan

The baySNP number refers to an internal numbering of the PA SNPs. Primer sequences are listed for amplification of the genomic fragments. In addition the respective fluorescent hybridisation probes are listed. If not otherwise stated, all fluorescent probes have a 'minor groove binder' (MGB) attached (Kutyavin et al., Nucleic Acids Research 2000, 28:655-661).

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baysNP	F-Sequence	R-Sequence	VIC-MGB	FAM-MGB
52	CACCCCTCTGAAATTCACTTAAATTCAA	GGCCCTTGAAAGAAGTTTATTTGAGAA	CTATGCCATACTTTTGC	AATGCTATAGTTTTCGATTAT
C				
542	TTCGCTCCATCAACCAAGTC	GATGGGTGATCAGCGGAATC	CAATTGGGAATTGGGAGGG	AATTGGGGTTGGGAGGG
821	GCCCAGTTATACCTCACTGTGTAAAC	AGGTCACTACAGAGGGTATCATGAGA	TGTGATACTCTGAAACAG	CTGTGATAACCTGGCACACA
1056	TGTATGGCACCTCGTGTATCTG	CGCTCTCGGCACTCTTG	CCAARACAaCAGGACGG	AAACAGCAGGACGGG
1204	CTGTAAGCTATCTGGATTGTCAATGA	GGCTCACTGCTTGTATCTTAAAG	CACTCACATTAAATTAG	ACTCACACATTAAATTAGT
1722	GGACCCCTAAGAACCCAGGAT	ATGGCTTAACACAGGAGATGATG	TGGCCTGGGGATGT	TGGCCTGGGGATGT
1757	ACAGGGCTGGCGCAC	AGCCTCTGCCCTCTCCA	AACCAAAATGAGGAGAG	ACCAAAATGAGGAGAG
1765	GGAGGCTTGAGCTAAGGGCTT	TGTCAGATGCACTGAAGGTG	ACGGAGGAAGGAGGT	ACGGAGGAAGGAAT
1799	TTCGGGGGGTTGTCATGACA	TGGZCATATGGGGGACTCT	AGTGTGATCATCCTACTTT	CAGTGTGATCCTGTCACT
1837	CACTCAGCCCTGCTTTC	CATCCTTGGGGCTTGGT	TGAGGGCTACATGA	TCATGGGGGTACAT
1870	CTGGCTCTGACCCCTGCT	GGAGGATGCCATCTCGAACAA	TGCTCTCCTTCTCACAC	CCTCTTTCACACCGA
1988	CCGTGGCTTGCTGGTGACT	CTACCTGTCGGTGCATCATC	TCCTATACCTGGGTGT	CTATACGTGGGTGTAT
2000	TTCTCACTCTGATATAAAACTCAGAC	CGATGAAACAGTTGGATAAGGTGT	TACTCATCTCTCTAAATTAC	CAAATATCTACTCATTTTC
CC				
2085	TCATTAACATCAGCTTATTCGACTGTA	TCAAGACACTGAAACTTAAGAATTC	TGTACCGAAAGAAA	TGTACCGAAAGAAA
AA				
2281	GCTGCATTTGGAGGAGACTGATC	CGGTAACTTAAAGAACGGATTC	CATACCACAAAAACCA	ACCAACAAACCCAGGTC
2298	TGCTTAGGGTTTCTGCTGATATT	GGC2ACCGTGTAGACTTGATCTAA	TCTATGGGCATTTC	TATCATGGCCCTTTCA
2357	GCGAAGTGTGCGAACCCAA	GGTTACGTTCTGCTCTTGATCCT	AAGACGAAAATGaaATC	AAGACGAAAATGgATC
4838	AAGATGACCTTATGGCTGAGATG	TCTCGAACATGACCTTGTAGCTGT	AAGGAATTGCCCTGCCT	AAGAAACTGCCCTGCC
5320	GGGGATATAGTACMAAACAGCCTG	CAACTTAATCACTACTCATGAAAGCA	AAGGAAAGCTGGATATG	AGGAAAGCTGGGTATGT

baySNP	F-Sequence	R-Sequence	VIC-MGB	FAM-MGB
TCT				
5717	GGCCCGTCTGGCT	AACCCACACCTTCAGTCTAGAAA	VIC-CCACCTCCCTtCTAGCCCTCAATTGC-TAMRA	Fam-CCCACCTCCCTtCTAGCCCTCAATTGC-Tamra
5959	ACCGAAACAATGCCAACCA	CAGTGAAACCAAGGGATGTC	VIC-CGAATGTGgCTGCCAGCC-TAMRA	Fam-TGGAATGTGgCTGCCAGCC-TAMRA
6482	CATAGTTAGGATAAACAAAGGATT	TGTCTATGAAACGCCAACAAC	AACGAACTGGTCTACCT	AGATCTGGTCTGCTC
8060	GCTATTGAATGGATTCAGCTTATT	TGCATGGATTCAGCATATGTT	CCC2ACCTGGAGAAT	TCCCACCTGGGGAA
8816	CAGCCCCCTCTGCTCCAG	TCCCCCTCTGCTCCAGGC	TGAGAAAAAAAGgtTCGG	CTGAGAAAAAAAGGTC
10600	GGTGAAGTTTGGCAATCTC	AAGTTAAATCAGCCCTTCAATTGG	TGCTTGGAAEAGCC	TGCTTGGAGACAGGC
10771	CTGGGCCACCGAGTTCAC	GATCTCTGAGTCAGTGGCTCT	AGGAAGGCTGGCCT	CAAGGAAGGTTGGC
10948	ACATCCCCCTTCCACGCTT	GCAGGGCAGAGGAGGA	CGCCCCAGTAATAACAGA	CCCAAGTAATCCAGACAC
11001	GGCATCCCTGTTGAACGTGAA	ACATGACCAGGGCCACTT	TCTTTCCACTGGACGT	TTCCATTGGACGTCT
11073	GAGCAACAGCCCTGAG	GCGGGAGCTAGAGGAGGTG	TCTGGCGCTGTC	TCTCGGGCTGTC
111248	GAAAGCTAACTCCCCTGACG	TGAAGGGTAAGGGAGGGAAA	CTTGGCGTCTGCGTC	TTGGCATCGCGTCAG
11654	AGTTTGTTCCTATAGGGTTCCA	CTCTTATGCCCTCCCCACCA	TTGAAAAGGACACCATATT	ACACCGTATTTTAC
11655	CATATTCAAGAAAGGATTATCTCCAACT	TGGAACACCTCTAAATAGGAAACAACT	CACTAAAGCTGTAAATATTA	CTAAAGCTGTATATTAC
CTT				
13191	GAAGTTGGCATAAAACCTAA	CCTGCCCCACCTTCTCTCT	TCTTCCCTCTGgtTACA	TCCCTCTGAGTAAACAC

Table 3 PA SNPs, SNP classes and putative PA genes

The baySNP number refers to an internal numbering of the PA SNPs. Listed are the different polymorphisms found in our association study. Also from the association study we defined SNP classes; with ADR being adverse drug reaction related, with EFF being drug efficacy related and CVD being cardiovascular disease related. ADR3 and ADR5 relate to advanced and severe ADR, whereas VEFF and UEFF relate to very high/low and ultra high/low drug efficacy (see table 1b). Also accession numbers and descriptions of those gene loci are given that are most homologous to the PA genes as listed in the sequences section (see below). Homologous genes and their accession numbers could be found by those skilled in the art in the Genbank database. Null: not defined.

BAYSNP	SNP class	GTYPE1	GTYPE1	GTYPE2	NCBI	DESCRIPTION
		1	2	2		
28	EFF	CC	CT	TT	U15552	Human acidic 82 kDa protein mRNA, complete cds.
29	CVD	AA	AG	GG	HS162961	Human T-lymphoma invasion and metastasis inducing TIAM1 protein (TIAM1) mRNA, complete cds.
29	ADR3	AA	AG	GG	HS162961	Human T-lymphoma invasion and metastasis inducing TIAM1 protein (TIAM1) mRNA, complete cds.
29	ADR5	AA	AG	GG	HS162961	Human T-lymphoma invasion and metastasis inducing TIAM1 protein (TIAM1) mRNA, complete cds.
52	EFF	CC	CG	GG	X69907	H.sapiens gene for mitochondrial ATP synthase c subunit (P1 form)

BAYSNP	SNP class	GTTYPE1	GTTYPE1	GTTYPE2	NCBI	DESCRIPTION
56	EFF	AA	AG	GG	M92357	Homo sapiens B94 protein mRNA, complete cds.
89	CVD	AA	AG	null	L23982	Homo sapiens (clones: CW52-2, CW27-6, CW15-2, CW26-5, 11-67) collagen type VII intergenic region and (COL7A1) gene, complete cds.
90	CVD	CC	CT	TT	M65212	Homo sapiens catechol-O-methyltransferase (COMT) mRNA, complete cds.
99	CVD	CC	CT	TT	X96698	H.sapiens mRNA for D1075-like gene
140	EFF	CC	CT	TT	M14335	Human coagulation factor V mRNA, complete cds.
152	EFF	AA	AG	GG	M32670	Homo sapiens ITGB3 gene, intron 2, fragment C, partial sequence.
214	CVD	AA	AG	GG	X66957	H. sapiens hexokinase I (MK-16)
221	CVD	CC	CG	GG	X76732	H.sapiens mRNA for NBRA protein
224	CVD	CC	CT	TT	M14764	Human nerve growth factor receptor mRNA, complete cds.
294	CVD	CC	CT	TT	P02568	ACTIN, ALPHA SKELETAL MUSCLE (ALPHA-ACTIN 1).
307	CVD	CC	CT	TT	X63346	H.sapiens mRNA for tre oncogene (clone 210)
411	CVD	AA	AT	TT	HS34804	Human thermostable phenol sulfotransferase (STP2) gene, partial cds.
449	CVD	CC	CG	GG	M36341	Human ADP-ribosylation factor 4 (ARF4) mRNA, complete cds.

BAY SNP	SNP class	GTYPE1 1	GTYPE1 2	GTYPE2 1	GTYPE2 2	NCBI	DESCRIPTION
466	CVD	CC	CT	TT	TT	AF129756	Homo sapiens MSH55 gene, partial cds; and CLIC1, DDAH, G6b, G6c, G5b, G6d, G6e, G6f, BAT5, G5b, CSK2B, BAT4, G4, Apo M, BAT3, BAT2, AIF-1, 1C7, LST-1, LTB, TNF, and LTA genes, complete cds.
472	EFF	AA	AG	GG	GG	M57965	Homo sapiens (clones lambda gMHC 1,2,3 and 4) beta-myosin heavy chain (MYH7) gene, complete cds.
542	CVD	AA	AG	GG	GG	M64082	Human flavin-containing monooxygenase (FMO1) mRNA, complete cds.
542	ADR	AA	AG	GG	GG	M64082	Human flavin-containing monooxygenase (FMO1) mRNA, complete cds.
739	CVD	CC	CG	GG	GG	I43509	Homo sapiens methionine adenosyltransferase alpha subunit gene fragment.
821	CVD	AA	AC	CC	CC	X80507	H.sapiens YAP65 mRNA
821	VEFF	AA	AC	CC	CC	X80507	H.sapiens YAP65 mRNA
1005	CVD	AA	AG	GG	GG	M81357	Human coagulation factor VII (F7) gene exon 1 and factor X (F10) gene, exon 1.
1055	CVD	AA	AT	TT	TT	J02758	Human apolipoprotein A-IV gene, complete cds.
1056	EFF	AA	AG	GG	GG	Q16720	CALCIUM-TRANSPORTING ATPASE PLASMA MEMBRANE, ISOFORMS 3A/3B (EC 3.6.1.38) (CALCIUM

BAYSNP	SNP class	GTTYPE1	GTTYPE1	GTTYPE2	GTTYPE2	NCBI	DESCRIPTION
		1	2	2	2		PUMP) (PMCA3).
1085	CVD	AA	AG	GG	GG	M14564	Human cytochrome P450c17 (steroid 17-alpha-hydroxylase/17,20 lyase) mRNA, complete cds.
1086	CVD	AA	AG	GG	GG	M14564	Human cytochrome P450c17 (steroid 17-alpha-hydroxylase/17,20 lyase) mRNA, complete cds.
1092	CVD	CC	CG	GG	GG	AF022375	Homo sapiens vascular endothelial growth factor mRNA, complete cds.
1096	CVD	GG	GT	TT	TT	X15323	H.sapiens angiotensinogen gene 5' region and exon 1
1101	EFF	CC	CT	TT	TT	AL031005	Homo sapiens DNA sequence from PAC 329E20 on chromosome 1p34.4-36.13. Contains endothelin-converting enzyme 1 (ECE-1), EST, STS, CA repeat
1204	CVD	AA	AG	GG	GG	AC004264	Homo sapiens PAC clone RP1-102K2 from 22q12.1-qter, complete sequence.
1504	CVD	CC	CT	TT	TT	AC005175	Homo sapiens chromosome 19, cosmid R31449, complete sequence.
1511	EFF	GG	GT	TT	TT	AF009674	Homo sapiens axin (AXIN) mRNA, partial cds.
1524	ADR3	AA	AC	CC	CC	AF223404	Homo sapiens WNT1 inducible signaling pathway protein 1 (WISP1) gene, promoter and partial cds.
1556	EFF	CC	CG	GG	GG	L34058	Homo sapiens cadherin-13 mRNA, complete cds.

BAYSNP	SNP class	GTTYPE1	GTTYPE2	NCBI	DESCRIPTION
1561	CVD	AA	AC	CC	M31664 Human cytochrome P450 (CYP1A2) gene, exons 1 and 2.
1582	CVD	CC	CT	TT	AF050163 Homo sapiens lipoprotein lipase precursor, gene, partial cds.
1638	CVD	AA	AG	GG	AF090318 Homo sapiens sterol 12-alpha hydroxylase CYP8B1 (Cyp8b1) mRNA, partial cds.
1653	CVD	GG	GT	TT	J02846 Human tissue factor gene, complete cds.
1662	CVD	CC	CT	TT	K02402 Human coagulation factor IX gene, complete cds.
1714	CVD	AA	AG	GG	D50857 Human DOCK180 protein mRNA, complete cds.
1722	ADR5	CC	CT	TT	D73409 Homo sapiens mRNA for diacylglycerol kinase delta, complete cds.
1757	EFF	AA	AG	GG	J04046 Human calmodulin mRNA, complete cds.
1765	ADR3	AA	AG	GG	J05096 Human Na,K-ATPase subunit alpha 2 (ATP1A2) gene, complete cds.
1765	ADR5	AA	AG	GG	J05096 Human Na,K-ATPase subunit alpha 2 (ATP1A2) gene, complete cds.
1776	CVD	AA	AG	GG	L22569 Homo sapiens cathepsin B mRNA, 3' UTR with a stem-loop structure providing mRNA stability.
1799	CVD	CC	CT	TT	D21255 Human mRNA for OB-cadherin-2, complete cds.
1806	EFF	AA	AG	GG	AF106202 Homo sapiens endothelial cell protein C receptor precursor

BAYSNP	SNP class	GTTYPE1	GTTYPE1	GTTYPE2	GTTYPE2	NCBI	DESCRIPTION
		1	2	1	2		(EPCR) gene, complete cds.
1837	CVD	CC	CT	TT	J00098		Human apolipoprotein A-I and C-III genes, complete cds.
1837	ADRS	CC	CT	TT	X00566		Human mRNA for lipoprotein apoAI Human apolipoprotein A-I and C-III genes, complete cds.
1837	ADR	CC	CT	TT	J00098		Human apolipoprotein A-I and C-III genes, complete cds.
1870	CVD	CC	CT	TT	M84820		Human retinoid X receptor beta (RXR-beta) mRNA, complete cds.
1882	CVD	CC	CT	TT	U06643		Human keratinocyte lectin 14 (HKL-14) mRNA, complete cds.
1988	CVD	CC	CT	TT	X61598		H.sapiens mRNA for collagen (a collagen-binding protein)
2000	CVD	CC	TT	null	P03915		NADH-UBIQUINONE OXIDOREDUCTASE CHAIN 5 (EC 1.6.5.3).
2000	ADR	CC	TT	null	P03915		NADH-UBIQUINONE OXIDOREDUCTASE CHAIN 5 (EC 1.6.5.3).
2071	CVD	AA	AG	GG	L04143		Human c-kit gene.
2078	CVD	GG	GT	TT	X77584		H.sapiens mRNA for ATL-derived factor/thioredoxin.
2085	VEFF	GG	GT	TT	X82540		H.sapiens mRNA for activin beta-C chain
2095	CVD	AG	GG	null	L34155		Homo sapiens laminin-related protein (LamA3) mRNA, complete cds.

BAYSNP	SNP class	GTYPE1	GTYPE1	GTYPE2	NCBI	DESCRIPTION
2119	CVD	AA	AG	null	Z22535	H.sapiens ALK-3 mRNA.
2119	EFF	AA	AG	null	Z22535	H.sapiens ALK-3 mRNA.
2141	EFF	AA	AG	GG	AB035073	Homo sapiens mRNA for platelet glycoprotein VI, complete cds.
2141	CVD	AA	AG	GG	AB035073	Homo sapiens mRNA for platelet glycoprotein VI, complete cds.
2182	EFF	AA	AG	GG	D32046	Human gene for thrombopoietin, exon1-exon6, complete cds.
2234	CVD	GG	GT	TT	AC004264	Homo sapiens PAC clone RP1-102K2 from 22q12.1-qter, complete sequence.
2281	VEFF	AA	AC	CC	X87872	H.sapiens mRNA for hepatocyte nuclear factor 4c
2298	CVD	AA	AC	CC	V01511	H.sapiens gene for beta-nerve growth factor (beta-NGF)
2341	CVD	CC	CT	TT	J03280	Human phenylethanolamine N-methyltransferase gene, complete cds.
2357	CVD	AA	AG	GG	O15055	PERIOD CIRCADIAN PROTEIN 2 (KIAA0347).
2366	CVD	GG	GT	TT	P35414	PROBABLE G PROTEIN-COUPLED RECEPTOR API.
2423	CVD	AA	AG	GG	AF000571	Homo sapiens kidney and cardiac voltage dependent K+ channel (KvLQT1) mRNA, complete cds.
2708	CVD	CC	CT	TT	AL031005	Homo sapiens DNA sequence from PAC 329E20 on chromosome 1p34.4-36.13. Contains endothelin-converting-enzyme 1 (ECE-1), EST, STS, CA repeat

BAYSNP	SNP class	GTYPE1	GTYPE2	NCBI	DESCRIPTION
2995	ADR5	AA	AC	CC	ABCC1: ATP-binding cassette, sub-family C (CFTR/MRP), member 1
2995	UEFF	AA	AC	CC	ABCC1: ATP-binding cassette, sub-family C (CFTR/MRP), member 1
3360	ADR5	GG	GT	TT	ABCB4: ATP-binding cassette, sub-family B (MDR/TAP), member 4
3464	CVD	AA	AG	GG	Human protein tyrosine phosphatase (PTPase-alpha) mRNA.
3689	EFF	CC	CG	GG	M34668 H.sapiens centromere autoantigen C (CENPC) mRNA, complete cds.
3975	UEFF	AA	AC	CC	U43368 Human VEGF related factor isoform VRF186 precursor (VRF) mRNA, complete cds.
3976	UEFF	AA	AG	GG	U43368 Human VEGF related factor isoform VRF186 precursor (VRF) mRNA, complete cds.
4206	ADR3	AA	AT	TT	BC000006 Homo sapiens, ATPase, Na+/K+ transporting, beta 1 polypeptide
4838	VEFF	AA	AG	GG	L08246 Human myeloid cell differentiation protein (MCL1) mRNA.
4912	EFF	AA	AG	GG	AF022375 Homo sapiens vascular endothelial growth factor mRNA, complete cds.
4925	CVD	AA	AC	CC	AF036365 Homo sapiens caveolin-3 (CAV3) mRNA, complete cds.

BAYSNP	SNP class	GTYPE1 1	GTYPE1 2	GTYPE2 1	GTYPE2 2	NCBI accession	DESCRIPTION
4966	ADR3	AA	AG	GG	GG	AF133298	Homo sapiens cytochrome P450 (CYP4F8) mRNA, complete cds.
5014	ADRS5	AA	AG	GG	GG	AL008637	Human DNA sequence from clone CTA-833B7 on chromosome 22q12.3-13.2. Contains the NCF4 gene for cytosolic neutrophil factor 4 (40kD), the 5' part of the CSF2RB gene for granulocyte-macrophage low-affinity colony stimulating factor 2 receptor beta, ESTs, STS
5296	CVD	AA	AG	GG	GG	J02933	Human blood coagulation factor VII gene, complete cds.
5296	EFF	AA	AG	GG	GG	J02933	Human blood coagulation factor VII gene, complete cds.
5298	EFF	CC	CT	TT	TT	J02933	Human blood coagulation factor VII gene, complete cds.
5298	CVD	CC	CT	TT	TT	J02933	Human blood coagulation factor VII gene, complete cds.
5320	EFF	AA	AG	GG	GG	J03799	Human colin carcinoma laminin-binding protein mRNA, complete cds.
5361	CVD	AA	AC	CC	CC	L02932	Human peroxisome proliferator activated receptor mRNA, complete cds.
5457	EFF	AA	AG	GG	GG	L29529	Homo sapiens (clone HHT-1 variant harboring HH-05) cardiac L-type voltage dependent calcium channel alpha 1 subunit (CACNL1A1) mRNA, complete cds.
5704	CVD	CC	CT	TT	TT	M58050	Human membrane cofactor protein (MCP) mRNA, complete cds.

BAYSNP	SNP class	GTTYPE1	GTTYPE1	GTTYPE2	GTTYPE2	NCHI	DESCRIPTION
		1	2	1	2		
5717	ADR3	AA	AG	GG	AL008637		Human DNA sequence from clone CTA-833B7 on chromosome 22q12.3-13.2 Contains the NCF4 gene for cytosolic neutrophil factor 4 (40kD), the 5' part of the CSF2RB gene for granulocyte-macrophage low-affinity colony stimulating factor 2 receptor beta, ESTs, STS
5959	CVD	AA	AG	GG	S	HSHMGCOA	H.sapiens mRNA for 3-hydroxy-3-methylglutaryl coenzyme A synthase
5959	ADR5	AA	AG	GG	S	HSHMGCOA	H.sapiens mRNA for 3-hydroxy-3-methylglutaryl coenzyme A synthase
5959	ADR	AA	AG	GG	S	HSHMGCOA	H.sapiens mRNA for 3-hydroxy-3-methylglutaryl coenzyme A synthase
6162	ADR3	CC	CG	GG	AF005896		Homo sapiens Na K-ATPase beta-3 subunit (atp1b3) gene, exon 7 and complete cds.
6162	ADR	CC	CG	GG	AF005896		Homo sapiens Na K-ATPase beta-3 subunit (atp1b3) gene, exon 7 and complete cds.
6162	ADR5	CC	CG	GG	AF005896		Homo sapiens Na K-ATPase beta-3 subunit (atp1b3) gene, exon 7 and complete cds.
6236	ADR5	CC	CT	TT	HSU62961		Human succinyl CoA:3-oxoacid CoA transferase precursor (OXCT) mRNA, complete cds.

BAYSNP	SNP class	GTYPE1	GTYPE2	NCBI	DESCRIPTION
6236	ADR3	CC	CT	TT	HSU62961 Human succinyl CoA:3-oxoacid transferase precursor (OXCT) mRNA, complete cds.
6482	CVD	AA	AG	GG	X69086 H.sapiens mRNA for utrophin
6498	CVD	AA	AG	GG	X71348 Homo sapiens vHNF1-C mRNA
6744	ADR5	CC	CT	TT	AC002310 Human Chromosome 16 BAC clone C1987SK-A-635H12, complete sequence.
7133	CVD	CC	CG	GG	K02402 Human coagulation factor IX gene, complete cds.
8021	CVD	AA	AG	GG	Z13009 H.sapiens mRNA for E-cadherin
8060	CVD	AA	AG	GG	Z99572 Human DNA sequence from PAC 86F14 on chromosome 1q23-1q24. Contains coagulation factor V, ESTs and STS.
8210	EFF	AA	AG	GG	ABCB11: ATP-binding cassette, sub-family B (MDR/TAP), member 11
8592	VEFF	CC	CT	TT	J04038 Human glyceraldehyde-3-phosphate dehydrogenase (GAPDH) gene, complete cds.
8816	EFF	CC	CG	GG	L36033 Human pre-B cell stimulating factor homologue (SDF1b) mRNA, complete cds.
8846	CVD	AA	AG	GG	L41162 Homo sapiens collagen alpha 3 type IX (COL9A3) mRNA, complete cds.

BAYSNP	SNP class	GTYPE1	GTYPE1	GTYPE2	NCBI	DESCRIPTION
8943	CVD	AA	AC	CC	AF050163	Homo sapiens lipoprotein lipase precursor, gene, partial cds.
9193	CVD	CC	CG	GG	M12674	Human estrogen receptor mRNA, complete cds.
9443	CVD	CC	CT	TT	U09587	Human glycyl-tRNA synthetase mRNA, complete cds.
9516	CVD	AA	AG	GG	U16720	Human interleukin 10 (IL10) gene, complete cds.
9698	ADR	AA	AG	GG	HS5211110	Homo sapiens X28 region near ALD locus containing dual specificity phosphatase 9 (DUSP9), ribosomal protein L18a (RPL18a), Ca2+/Calmodulin-dependent protein kinase I (CAMK1), creatine transporter (CRTR), CDM protein (CDM), adrenoleukodystrophy protein (AL)
9698	ADR3	AA	AG	GG	HS5211110	Homo sapiens X28 region near ALD locus containing dual specificity phosphatase 9 (DUSP9), ribosomal protein L18a (RPL18a), Ca2+/Calmodulin-dependent protein kinase I (CAMK1), creatine transporter (CRTR), CDM protein (CDM), adrenoleukodystrophy protein (AL)
9698	EFF	AA	AG	GG	HS5211110	Homo sapiens X28 region near ALD locus containing dual specificity phosphatase 9 (DUSP9), ribosomal protein L18a (RPL18a), Ca2+/Calmodulin-dependent protein kinase I (CAMK1), creatine transporter (CRTR), CDM protein (CDM), adrenoleukodystrophy protein (AL)

BAYSNP	SNP class	GTTYPE1	GTTYPE1	GTTYPE2	GTTYPE2	NCBI	DESCRIPTION
9698	ADRS	AA	AG	GG	GG	HS5211110	Homo sapiens X28 region near ALD locus containing dual specificity phosphatase 9 (DUSP9), ribosomal protein L18a (RPL18a), Ca2+/Calmodulin-dependent protein kinase I (CAMK1), creatine transporter (CRTTR), CDM protein (CDM), adrenoleukodystrophy protein (AL)
9698	CVD	AA	AG	GG	GG	HS5211110	Homo sapiens X28 region near ALD locus containing dual specificity phosphatase 9 (DUSP9), ribosomal protein L18a (RPL18a), Ca2+/Calmodulin-dependent protein kinase I (CAMK1), creatine transporter (CRTTR), CDM protein (CDM), adrenoleukodystrophy protein (AL)
9849	CVD	CC	CT	null	X04588		Human 2.5 kb mRNA for cytoskeletal tropomyosin TIV30(nm)
9883	CVD	AA	AG	GG	BC000140	PCCA: propionyl Coenzyme A carboxylase, alpha polypeptide	
10079	CVD	AA	AG	GG	X77197	H.sapiens mRNA for chloride channel	
10481	ADRS	AA	AT	TT	AF023268	Homo sapiens clk2 kinase (CLK2), propin1, cotel, glucocerebrosidase (GBA), and metaxin genes, complete cds; metaxin pseudogene and glucocerebrosidase pseudogene; and thrombospondin3 (THBS3) gene, partial cds.	
10542	UEFF	CC	CT	TT	AF066859	Homo sapiens muscle glycogen phosphorylase (PYGM) mRNA, complete cds.	

BAYSNP	SNP class	GTYPE1	GTYPE1	GTYPE2	NCBI	DESCRIPTION
		1	2	2		
10542	ADRS	CC	CT	TT	AF066859	Homo sapiens muscle glycogen phosphorylase (PYGM) mRNA, complete cds.
10600	EFF	AA	AG	GG	AF129756	Homo sapiens MSH55 gene, partial cds; and CLIC1, DDAH, G6b, G6c, G5b, G6d, G6e, G6f, BAT5, G5b, CSK2B, BAT4, G4, Apo M, BAT3, BAT2, AIF-1, IC7, LST-1, LTB, TNF, and LTA genes, complete cds.
10621	CVD	CC	CT	TT	AF220490	Homo sapiens group III secreted phospholipase A2 mRNA, complete cds.
10745	ADRS	AA	AG	GG	D11456	Human mRNA for Xanthine dehydrogenase, complete cds.
10745	VEFF	AA	AG	GG	D11456	Human mRNA for Xanthine dehydrogenase, complete cds.
10747	ADR	CC	CT	TT	D11456	Human mRNA for Xanthine dehydrogenase, complete cds.
10747	CVD	CC	CT	TT	D11456	Human mRNA for Xanthine dehydrogenase, complete cds.
10747	ADR3	CC	CT	TT	D11456	Human mRNA for Xanthine dehydrogenase, complete cds.
10771	ADRS	CC	CG	GG	D37932	Human mRNA for HPC-1, partial cds.
10771	EFF	CC	CG	GG	D37932	Human mRNA for HPC-1, partial cds.
10870	CVD	AA	AG	GG	AH002776	LDLR: low density lipoprotein receptor (familial hypercholesterolemia)
10877	CVD	AA	AC	CC	AC005832	Homo sapiens 12p13.3 BAC RPCI11-500M8 (Roswell Park

BAY SNP	SNP class	GTTYPE1	GTTYPE2	NCBI	DESCRIPTION
		1	2		
					Cancer Institute Human BAC Library) complete sequence.
10948	CVD	GG	GT	TT	M10065 Human apolipoprotein E (epsilon-4 allele) gene, complete cds.
11001	ADR5	CC	CT	TT	M34424 Human acid alpha-glucosidase (GAA) mRNA, complete cds.
11073	ADR5	CC	CG	GG	AF070670 Homo sapiens protein phosphatase 2C alpha 2 mRNA, complete cds.
11153	CVD	CC	CT	TT	U57623 Human fatty acid binding protein FABP gene, complete cds.
11210	CVD	CC	CT	TT	AB014460 Homo sapiens TSC2, NTHL1/NTH1 and SLC9A3R2/E3KARP genes, partial and complete cds.
11210	ADR3	CC	CT	TT	AB014460 Homo sapiens TSC2, NTHL1/NTH1 and SLC9A3R2/E3KARP genes, partial and complete cds.
11210	ADR	CC	CT	TT	AB014460 Homo sapiens TSC2, NTHL1/NTH1 and SLC9A3R2/E3KARP genes, partial and complete cds.
11248	ADR	CC	CT	TT	X60435 H.sapiens gene PACAP for pituitary adenylate cyclase activating polypeptide
11248	CVD	CC	CT	TT	X60435 H.sapiens gene PACAP for pituitary adenylate cyclase activating polypeptide
11372	CVD	AA	AG	GG	Z82215 Human DNA sequence from clone RP1-68O2 on chromosome 22 Contains the 5' end of the APOL2 gene for apolipoprotein L2, the APOL gene for apolipoprotein L, the MYH9 gene for

BAYSNP	SNP class	GTTYPE1 1	GTTYPE1 2	GTTYPE2 1	GTTYPE2 2	NCBI	DESCRIPTION
11449	CVD	CC	CG	GG	GG	AF050163	Homo sapiens lipoprotein lipase precursor, gene, partial cds.
11450	EFF	AA	AT	TT	TT	AF050163	Homo sapiens lipoprotein lipase precursor, gene, partial cds.
11470	CVD	CC	CT	null	AJ006945	Human P2Y1 gene	
11472	CVD	AA	AT	null	AJ006945	Human P2Y1 gene	
11487	ADRS	AT	TT	null	M75106	Human prepro-plasma carboxypeptidase B mRNA, complete cds.	
11487	ADR3	AT	TT	null	M75106	Human prepro-plasma carboxypeptidase B mRNA, complete cds.	
11488	ADRS	CC	CG	GG	M75106	Human prepro-plasma carboxypeptidase B mRNA, complete cds.	
11488	UEFF	CC	CG	GG	M75106	Human prepro-plasma carboxypeptidase B mRNA, complete cds.	
11488	ADR3	CC	CG	GG	M75106	Human prepro-plasma carboxypeptidase B mRNA, complete cds.	
11493	CVD	AA	AG	GG	U03882	Human monocyte chemoattractant protein 1 receptor (MCP-1RA) alternatively spliced mRNA, complete cds.	
11502	ADR3	CC	CT	TT	U58917	Homo sapiens IL-17 receptor mRNA, complete cds.	
11502	ADR5	CC	CT	TT	U58917	Homo sapiens IL-17 receptor mRNA, complete cds.	
11534	CVD	GG	GT	null	AJ276102	Homo sapiens mRNA for GPRC5C protein	

BAYSNP	SNP class	GTTYPE1	GTTYPE1	GTTYPE2	GTTYPE2	NCBI	DESCRIPTION
11537	CVD	AA	AG	GG	GG	AL022721	Human DNA sequence from clone 109F14 on chromosome 6p21.2-21.3. Contains the alternatively spliced gene for Transcriptional Enhancer Factor TEF-5, the 60S Ribosomal Protein RPL10A gene, a PUTATIVE ZNF127 LIKE gene, and the PPARD for Peroxisome Proliferator
11537	EFF	AA	AG	GG	GG	AL022721	Human DNA sequence from clone 109F14 on chromosome 6p21.2-21.3. Contains the alternatively spliced gene for Transcriptional Enhancer Factor TEF-5, the 60S Ribosomal Protein RPL10A gene, a PUTATIVE ZNF127 LIKE gene, and the PPARD for Peroxisome Proliferator
11560	EFF	AA	AG	GG	GG	AC006312	Homo sapiens chromosome 9, clone hRPK.401_G_18, complete sequence.
11578	CVD	CC	CT	CT	null	AC073593	Homo sapiens 12 BAC RP11-13J12 (Roswell Park Cancer Institute Human BAC Library) complete sequence.
11594	ADR3	CC	CT	TT	TT	AF026069	Homo sapiens phosphomevalonate kinase (HUMPMK1) gene, partial cds.
11594	ADR5	CC	CT	TT	TT	AF026069	Homo sapiens phosphomevalonate kinase (HUMPMK1) gene, partial cds.
11594	CVD	CC	CT	TT	TT	AF026069	Homo sapiens phosphomevalonate kinase (HUMPMK1) gene,

BAY SNP	SNP class	GTTYPE1	GTTYPE1	GTTYPE2	GTTYPE2	NCBI	DESCRIPTION
		1	2	1	2		
11594	ADR	CC	CT	TT	AF026069	Homo sapiens phosphomevalonate kinase (HUMPMK1) gene, partial cds.	partial cds.
11624	CVD	CC	CT	TT	AL022721	Human DNA sequence from clone 109F14 on chromosome 6p21.2-21.3. Contains the alternatively spliced gene for Transcriptional Enhancer Factor TEF-5, the 60S Ribosomal Protein RPL10A gene, a PUTATIVE ZNF127 LIKE gene, and the PPARD for Peroxisome Proliferato	Human DNA sequence from clone 109F14 on chromosome 6p21.2-21.3. Contains the alternatively spliced gene for Transcriptional Enhancer Factor TEF-5, the 60S Ribosomal Protein RPL10A gene, a PUTATIVE ZNF127 LIKE gene, and the PPARD for Peroxisome Proliferato
11624	EFF	CC	CT	TT	AL022721	Human DNA sequence from clone 109F14 on chromosome 6p21.2-21.3. Contains the alternatively spliced gene for Transcriptional Enhancer Factor TEF-5, the 60S Ribosomal Protein RPL10A gene, a PUTATIVE ZNF127 LIKE gene, and the PPARD for Peroxisome Proliferato	Human DNA sequence from clone 109F14 on chromosome 6p21.2-21.3. Contains the alternatively spliced gene for Transcriptional Enhancer Factor TEF-5, the 60S Ribosomal Protein RPL10A gene, a PUTATIVE ZNF127 LIKE gene, and the PPARD for Peroxisome Proliferato
11627	CVD	CC	CT	TT	AL022721	Human DNA sequence from clone 109F14 on chromosome 6p21.2-21.3. Contains the alternatively spliced gene for Transcriptional Enhancer Factor TEF-5, the 60S Ribosomal Protein RPL10A gene, a PUTATIVE ZNF127 LIKE gene, and the PPARD for Peroxisome Proliferato	Human DNA sequence from clone 109F14 on chromosome 6p21.2-21.3. Contains the alternatively spliced gene for Transcriptional Enhancer Factor TEF-5, the 60S Ribosomal Protein RPL10A gene, a PUTATIVE ZNF127 LIKE gene, and the PPARD for Peroxisome Proliferato
11627	EFF	CC	CT	TT	AL022721	Human DNA sequence from clone 109F14 on chromosome 6p21.2-21.3. Contains the alternatively spliced gene for Transcriptional Enhancer Factor TEF-5, the 60S Ribosomal Protein RPL10A gene, a PUTATIVE ZNF127 LIKE gene, and the PPARD for Peroxisome Proliferato	Human DNA sequence from clone 109F14 on chromosome 6p21.2-21.3. Contains the alternatively spliced gene for Transcriptional Enhancer Factor TEF-5, the 60S Ribosomal Protein RPL10A gene, a PUTATIVE ZNF127 LIKE gene, and the PPARD for Peroxisome Proliferato

BAYSNP	SNP class	GTYPE1 1	GTYPE1 2	GTYPE2 1	GTYPE2 2	NCBI	DESCRIPTION
							6p21.2-21.3. Contains the alternatively spliced gene for Transcriptional Enhancer Factor TEF-5, the 60S Ribosomal Protein RPL10A gene, a PUTATIVE ZNF127 LIKE gene, and the PPARD for Peroxisome Proliferator
11644	ADRS	AA	AG	GG	GG	D84371	Homo sapiens mRNA for serum arylalkylphosphatase, complete cds.
11650	EFF	AA	AG	GG	GG	X56668	Human DNA for calretinin exon 1
11654	ADRS	AA	AG	GG	GG	AJ276180	Homo sapiens partial ZNF202 gene for zinc finger protein homolog, exon 4
11654	ADR3	AA	AG	GG	GG	AJ276180	Homo sapiens partial ZNF202 gene for zinc finger protein homolog, exon 4
11655	ADRS	AA	AC	CC	CC	AJ276180	Homo sapiens partial ZNF202 gene for zinc finger protein homolog, exon 4
11655	ADR3	AA	AC	CC	CC	AJ276180	Homo sapiens partial ZNF202 gene for zinc finger protein homolog, exon 4
11656	CVD	CC	CT	TT	TT	NM_001081	CUBN: cubilin (intrinsic factor-cobalamin receptor)
11656	EFF	CC	CT	TT	TT	NM_001081	CUBN: cubilin (intrinsic factor-cobalamin receptor)
11825	ADRS	AA	AG	null	null	AC008897	Homo sapiens chromosome 5 clone CTD-2235C13, WORKING DRAFT SEQUENCE, 6 ordered pieces.

BAYSNP	SNP class	GTTYPE1	GTTYPE1	GTTYPE2	GTTYPE2	NCBI	DESCRIPTION
		1	2	1	2		
11914	ADRS	AA	AT	TT		AF030555	Homo sapiens acyl-CoA synthetase 4 (ACS4) mRNA, complete cds.
12008	EFF	CC	CT	null		AF107885	Homo sapiens transforming growth factor-beta 3 (TGF-beta 3) gene, complete cds; and unknown genes.
12008	ADRS	CC	CT	null		AF107885	Homo sapiens transforming growth factor-beta 3 (TGF-beta 3) gene, complete cds; and unknown genes.
12097	ADRS	AG	GG	null		AF280107	Homo sapiens cytochrome P450 polypeptide 43 (CYP3A43) gene, partial cds; cytochrome P450 polypeptide 4 (CYP3A4) and cytochrome P450 polypeptide 7 (CYP3A7) genes, complete cds; and cytochrome P450 polypeptide 5 (CYP3A5) gene, partial cds.
12097	ADRS	AG	GG	null		AF280107	Homo sapiens cytochrome P450 polypeptide 43 (CYP3A43) gene, partial cds; cytochrome P450 polypeptide 4 (CYP3A4) and cytochrome P450 polypeptide 7 (CYP3A7) genes, complete cds; and cytochrome P450 polypeptide 5 (CYP3A5) gene, partial cds.
12366	UEFF	AA	AG	GG		D63807	Human mRNA for lanosterol synthase, complete cds.
12366	ADRS	AA	AG	GG		D63807	Human mRNA for lanosterol synthase, complete cds.
12619	ADRS	AG	GG	null		L13744	Human AF-9 mRNA, complete cds.

Table 4 Cohorts

Given are names (as used in table 5) and formations of the various cohorts that were used for genotyping

COHORT	Definition
HELD_ALL_GOOD/BAD	Healthy elderly individuals of both genders with good or bad serum lipid profiles (as defined in table 1a)
HELD_FEM_GOOD/BAD	Healthy elderly individuals (female) with good or bad serum lipid profiles (as defined in table 1a)
HELD_MAL_GOOD/BAD	Healthy elderly individuals (male) with good or bad serum lipid profiles (as defined in table 1a)
CVD_ALL_CASE/CTRL	Individuals with diagnosis of cardiovascular disease and healthy controls (both genders)
CVD_FEM_CASE/CTRL	Individuals with diagnosis of cardiovascular disease and healthy controls (female)
CVD_MAL_CASE/CTRL	Individuals with diagnosis of cardiovascular disease and healthy controls (male)
HELD_FEM_ADRCTRL	Female individuals that tolerate administration of cerivastatin without exhibiting signs of ADR (as defined in table 1b)
HELD_FEM_ADRCASE	Female individuals that exhibited ADR (as defined in table 1b) upon administration of cerivastatin
HELD_MAL_ADRCTRL	Male individuals that tolerate administration of cerivastatin without exhibiting signs of ADR (as defined in table 1b)
HELD_MAL_ADRCASE	Male individuals that exhibited ADR (as defined in table 1b) upon administration of cerivastatin
HELD_ALL_ADRCTRL	Individuals of both genders that tolerate administration of cerivastatin without exhibiting signs

COHORT	Definition
HELD_ALL_ADRCASE	of ADR (as defined in table 1b)
HELD_FEM_LORESP	Individuals of both genders that exhibited ADR (as defined in table 1b) upon administration of cerivastatin
HELD_FEM_HIRESP	Female individuals with a minor response to cerivastatin administration (as defined in table 1b)
HELD_FEM_HIHDL/LOHDL	Female individuals with a high response to cerivastatin administration (as defined in table 1b)
HELD_MAL_HIHDL/LOHDL	Healthy elderly individuals (female) with high or low serum HDL cholesterol levels (as defined in table 1c)
HELD_ALL_HIHDL/LOHDL	Healthy elderly individuals (male) with high or low serum HDL cholesterol levels (as defined in table 1c)
HELD_FEM_ADR3CASE	Female individuals of both genders with high or low serum HDL cholesterol levels (as defined in table 1c)
HELD_MAL_ADR3CASE	Female individuals that exhibited advanced ADR (as defined in table 1b) upon administration of cerivastatin
HELD_ALL_ADR3CASE	Male individuals that exhibited advanced ADR (as defined in table 1b) upon administration of cerivastatin
HELD_FEM_VLORESP	Individuals of both genders that exhibited advanced ADR (as defined in table 1b) upon administration of cerivastatin
	Female individuals with a very low response to cerivastatin administration (as defined in table 1b)

COHORT	Definition
HELD_FEM_VHIRESP	Female individuals with a very high response to cerivastatin administration (as defined in table 1b)
HELD_FEM_ADDR5CASE	Female individuals that exhibited severe ADR (as defined in table 1b) upon administration of cerivastatin
HELD_MAL_ADDR5CASE	Male individuals that exhibited severe ADR (as defined in table 1b) upon administration of cerivastatin
HELD_ALL_ADDR5CASE	Individuals of both genders that exhibited severe ADR (as defined in table 1b) upon administration of cerivastatin
HELD_FEM_ULORESP	Female individuals with a ultra low response to cerivastatin administration (as defined in table 1b)
HELD_FEM_UHRESP	Female individuals with a ultra high response to cerivastatin administration (as defined in table 1b)

Table 5a and 5b Cohort sizes and p-values of PA SNPs

The baySNP number refers to an internal numbering of the PA SNPs. Cpvval denotes the classical Pearson chi-squared test, Xpval denotes the likelihood-ratio chi-squared test, Cpvvalue, Xpvalue, and LRpvalue are calculated exact version of Pearson's chi-squared test, LRpval denotes the likelihood-ratio chi-squared test., Cpvval, Xpvalue, and LRpvalue are calculated as described in (SAS/STAT User's Guide of the SAS OnlineDoc, Version 8), (L. D. Fisher and G. van Belle, Biostatistics, Wiley Interscience 1993), and (A. Agresti, Statistical Science 7, 131 (1992)). The GTYPE and Allele p values were obtained through the respective chi square tests when comparing COHORTs A and B. For GTYPE p value the number of patients in cohort A carrying genotypes 11, 12 or 22 (FQ11 A, FQ12 A, FQ22 A; genotypes as defined in table 3) were compared with the respective patients in cohort B (FQ11 B, FQ12 B, FQ22 B; genotypes as defined in table 3) resulting in the respective chi square test with a 3x2 matrix. For Allele p values we compared the allele count of alleles 1 and 2 (A1 and A2) in cohorts A and B, respectively (chi square test with a 2x2 matrix). SIZE A and B: Number of patients in cohorts A and B, respectively. See table 4 for definition of COHORTs A and B.

Table 5a Cohort sizes and frequency of alleles and genotypes

baySNP		COHORT A	SIZE A	COHORT B		SIZE B	FQ1 B	FQ2 B	FQ1	FQ2	FQ12	FQ22
				A1	A2							
28	C T	HELD_FEM_HRESP	12	4	20	1	2	9	HELD_FEM_LORESP	22	18	26
29	A G	HELD_ALL_LHDL	10	12	8	4	4	2	HELD_ALL_HHDL	15	7	23
29	A G	HELD_MAL_ADRCTRL	26	33	19	13	7	6	HELD_MAL_ADRCTRL	72	68	76
29	A G	ULN							HELD_MAL_ADRCTRL	72	68	76
52	C G	HELD_MAL_ADRCASE5	9	13	5	5	3	1	HELD_MAL_ADRCTRL	72	68	76
		ULN							HELD_FEM_LORESP	31	27	35
		HELD_FEM_HRESP	18	24	12	7	10	1	HELD_FEM_LORESP	31	27	35
										5	17	9

baySNP	A1	A2	COHORT A		SIZE	FQ1 A		FQ2 A		FQ12		COHORT B		SIZE	FQ1 B		FQ2 B		FQ11		FQ12		FQ22	
			A	A		A	A	A	A	A	A	B	B		B	B	B	B	B	B	B	B	B	
56	A	G	HELD_FEM_HIRESP	12	5	19	0	5	7	HELD_FEM_LORESP	22	2	42	0	2	20								
89	A	G	HELD_ALL_CASE	45	90	0	45	0	0	HELD_ALL_CTRL	40	77	3	37	3	0								
90	C	T	HELD_FEM_CASE	31	29	33	8	13	10	HELD_FEM_CTRL	22	27	17	6	15	1								
99	C	T	HELD_FEM_BAD	82	54	110	13	28	41	HELD_FEM_GOOD	80	51	109	5	41	34								
140	C	T	HELD_FEM_HIRESP	12	24	0	0	0	12	HELD_FEM_LORESP	21	4	38	1	2	18								
152	A	G	HELD_FEM_HIRESP	12	12	12	3	6	3	HELD_FEM_LORESP	22	33	11	12	9	1								
214	A	G	HELD_ALL_BAD	97	156	38	59	38	0	HELD_ALL_GOOD	113	182	44	73	36	4								
214	A	G	HELD_FEM_BAD	81	131	31	50	31	0	HELD_FEM_GOOD	78	122	34	48	26	4								
221	C	G	HELD_ALL_CASE	45	26	64	7	12	26	HELD_ALL_CTRL	39	27	51	3	21	15								
221	C	G	HELD_FEM_CASE	31	17	45	4	9	18	HELD_FEM_CTRL	22	18	26	2	14	6								
224	C	T	HELD_FEM_BAD	79	110	48	51	8	20	HELD_FEM_GOOD	79	125	33	60	5	14								
224	C	T	HELD_MAL_BAD	20	35	5	17	1	2	HELD_MAL_GOOD	37	51	23	25	1	11								
294	C	T	HELD_ALL_CASE	45	56	34	16	24	5	HELD_ALL_CTRL	40	58	22	18	22	0								
307	C	T	CVD_FEM_CASE	36	19	53	2	15	19	CVD_FEM_CTRL	38	38	38	9	20	9								
307	C	T	HELD_ALL_BAD	102	70	134	0	70	32	HELD_ALL_GOOD	117	63	171	0	63	54								
411	A	T	HELD_ALL_LHDL	10	17	3	7	3	0	HELD_ALL_HHDL	15	18	12	5	8	2								
449	C	G	HELD_MAL_BAD	20	3	37	0	3	17	HELD_MAL_GOOD	37	16	58	1	14	22								
466	C	T	CVD_FEM_CASE	35	27	43	6	15	14	CVD_FEM_CTRL	40	44	36	12	20	8								
472	A	G	HELD_FEM_HIRESP	11	22	0	0	0	11	HELD_FEM_LORESP	22	12	32	3	6	13								
542	A	G	HELD_MAL_CASE	14	12	16	2	8	4	HELD_MAL_CTRL	19	2	36	0	2	17								
542	A	G	HELD_MAL_LHDL	21	14	28	3	8	10	HELD_MAL_HHDL	27	3	51	0	3	24								
542	A	G	HELD_ALL_ADRCASE	159	53	265	0	53	106	HELD_ALL_ADRCTRL	154	37	271	2	33	119								

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baySNP	A1	A2	COHORT_A	SIZE	FQ1_A	FQ2_A	FQ11	FQ12	FQ22	COHORT_B	SIZE	FQ1_B	FQ2_B	FQ11	FQ12	FQ22
542	A	G	HELD_FEM_LHDL	23	2	44	0	2	21	HELD_FEM_HHDL	32	10	54	1	8	23
739	C	G	HELD_ALL_CASE	45	39	51	9	21	15	HELD_ALL_CTRL	40	48	32	14	20	6
821	A	C	HELD_MAL_BAD2	309	180	438	32	116	161	HELD_MAL_GOOD2	349	174	524	18	138	193
821	A	C	HELD_FEM_VHRESP	10	4	16	0	4	6	HELD_FEM_VLORESP	14	14	14	4	6	4
1005	A	G	HELD_MAL_CASE	14	26	2	12	2	0	HELD_MAL_CTRL	18	27	9	11	5	2
1055	A	T	HELD_MAL_CASE	9	3	15	0	3	6	HELD_MAL_CTRL	12	8	16	4	0	8
1056	A	G	HELD_FEM_HIRESP	24	30	18	12	6	6	HELD_FEM_LORESP	33	41	25	10	21	2
1085	A	G	HELD_MAL_BAD	20	17	23	3	11	6	HELD_MAL_GOOD	36	46	26	15	16	5
1085	A	G	CVD_FEM_CASE	34	51	17	20	11	3	CVD_FEM_CTRL	40	47	33	16	15	9
1086	A	G	HELD_MAL_BAD	20	24	16	7	10	3	HELD_MAL_GOOD	36	28	44	5	18	13
1092	C	G	HELD_MAL_BAD	20	9	31	2	5	13	HELD_MAL_GOOD	37	29	45	4	21	12
1096	G	T	HELD_MAL_CASE	14	7	21	0	7	7	HELD_MAL_CTRL	18	3	33	0	3	15
1096	G	T	CVD_MAL_CASE	69	21	117	4	13	52	CVD_MAL_CTRL	33	12	54	0	12	21
1101	C	T	HELD_FEM_HIRESP	12	24	0	12	0	0	HELD_FEM_LORESP	22	40	4	18	4	0
1204	A	G	HELD_MAL_BAD	19	12	26	2	8	9	HELD_MAL_GOOD	35	9	61	0	9	26
1204	A	G	HELD_ALL_BAD	99	62	136	12	38	49	HELD_ALL_GOOD	115	52	178	8	36	71
1504	C	T	HELD_ALL_CASE	44	37	51	5	27	12	HELD_ALL_CTRL	39	36	42	12	12	15
1504	C	T	HELD_MAL_BAD	19	12	26	0	12	7	HELD_MAL_GOOD	37	33	41	8	17	12
1504	C	T	HELD_MAL_CASE	14	13	15	2	9	3	HELD_MAL_CTRL	18	12	24	4	4	10
1511	G	T	HELD_FEM_HIRESP	12	15	9	3	9	0	HELD_FEM_LORESP	22	35	9	14	7	1
1524	A	C	HELD_FEM_ADRCASE3	38	16	60	0	16	22	HELD_FEM_ADRCTRL	82	39	125	8	23	51

hapSNP	A1	A2	COHORT A	SIZE	FQ1 A	FQ2 A	FQ11	FQ12	FQ22	COHORT B	SIZE	FQ1 B	FQ2 B	FQ11	FQ12	FQ22
				A	A	A	A	A	A	B	B	B	B	B	B	B
			ULN													
1556	C	G	HLD_FEM_HIRESP	12	7	17	0	7	5	HELD_FEM_LORESP	22	3	41	0	3	19
1561	A	C	CVD_FEM_CASE	36	58	14	23	12	1	CVD_FEM_CTRL	40	53	27	17	19	4
1582	C	T	HLD_MAL_BAD	20	5	35	0	5	15	HELD_MAL_GOOD	37	22	52	5	12	20
1638	A	G	HLD_FEM_CASE	31	10	52	1	8	22	HELD_FEM_CTRL	22	15	29	2	11	9
1653	G	T	CVD_MAL_CASE	69	70	68	15	40	14	CVD_MAL_CTRL	33	30	36	10	10	13
1662	C	T	HLD_MAL_CASE	14	8	20	4	0	10	HELD_MAL_CTRL	18	36	0	0	0	18
1714	A	G	CVD_MAL_CASE	66	32	100	3	26	37	CVD_MAL_CTRL	34	26	42	6	14	14
1722	C	T	HLD_FEM_ADRCASES	18	21	15	8	5	5	HELD_FEM_ADRCTRL	81	71	91	14	43	24
			ULN													
1757	A	G	HLD_FEM_HIRESP	20	15	25	4	7	9	HELD_FEM_LORESP	32	16	48	0	16	16
1765	A	G	HLD_ALL_ADRCASE3	63	9	117	1	7	55	HELD_ALL_ADRCTRL	149	56	242	4	48	97
			ULN													
1765	A	G	HLD_ALL_ADRCASE3	63	9	117	1	7	55	HELD_ALL_ADRCTRL	149	56	242	4	48	97
1765	A	G	HLD_ALL_ADRCASES	27	3	51	0	3	24	HELD_ALL_ADRCTRL	149	56	242	4	48	97
1765	A	G	HLD_ALL_ADRCASES	27	3	51	0	3	24	HELD_ALL_ADRCTRL	149	56	242	4	48	97
1765	A	G	HLD_MAL_ADRCASE3	26	2	50	0	2	24	HELD_MAL_ADRCTRL	70	25	115	2	21	47
1765	A	G	HLD_MAL_ADRCASE3	26	2	50	0	2	24	HELD_MAL_ADRCTRL	70	25	115	2	21	47

baySNP	A1	A2	COHORT_A	SIZE	FQ1_A	FQ2_A	FQ11	FQ12	FQ22	COHORT_B	SIZE	FQ1_B	FQ2_B	FQ11	FQ12	FQ22
			A	A	A	A	A	A	A	B	B	B	B	B	B	B
			ULN													
1765	A	G	HELD_MAL_ADRCASE5	10	20	0	0	0	10	HELD_MAL_ADRCTRL	70	25	115	2	21	47
1765	A	G	HELD_MAL_ADRCASE5	10	20	0	0	0	10	HELD_MAL_ADRCTRL	70	25	115	2	21	47
1765	A	G	HELD_FEM_ADRCASE3	37	7	67	1	5	31	HELD_FEM_ADRCTRL	79	31	127	2	27	50
1765	A	G	HELD_FEM_ADRCASE3	37	7	67	1	5	31	HELD_FEM_ADRCTRL	79	31	127	2	27	50
1776	A	G	HELD_ALL_CASE	45	90	0	45	0	0	HELD_ALL_CTRL	40	74	6	37	0	3
1776	A	G	HELD_FEM_CASE	31	62	0	31	0	0	HELD_FEM_CTRL	22	40	4	20	0	2
1799	C	T	HELD_FEM_BAD2	291	365	217	123	119	49	HELD_FEM_GOOD2	356	468	244	145	178	33
1799	C	T	HELD_MAL_CASE	14	15	13	4	7	3	HELD_MAL_CTRL	18	28	8	11	6	1
1806	A	G	HELD_FEM_HRESP	12	23	1	11	1	0	HELD_FEM_LORESP	22	34	10	14	6	2
1837	C	T	HELD_FEM_BAD2	304	436	172	164	108	32	HELD_FEM_GOOD2	355	499	211	166	167	22
1837	C	T	HELD_ALL_BAD2	607	891	323	334	223	50	HELD_ALL_GOOD2	682	952	412	322	308	52
1837	C	T	HELD_ALL_ADRCASE5	28	46	10	20	6	2	HELD_ALL_ADRCTRL	155	208	102	66	76	13
1837	C	T	HELD_MAL_ADRCASE	77	107	47	37	33	7	HELD_MAL_ADRCTRL	72	86	58	21	44	7
1837	C	T	HELD_MAL_BAD2	303	455	151	170	115	18	HELD_MAL_GOOD2	327	453	201	156	141	30
1870	C	T	HELD_ALL_CASE	45	29	61	2	25	18	HELD_ALL_CTRL	39	16	62	3	10	26
1870	C	T	HELD_FEM_CASE	31	22	40	1	20	10	HELD_FEM_CTRL	22	9	35	1	7	14

baySNP	A1	A2	COHORT_A	SIZE	FQ1_A	FQ2_A	FQ11_A	FQ12_A	FQ22	COHORT_B	SIZE	FQ1_B	FQ2_B	FQ11_B	FQ12_B	FQ22_B
1882	C	T	CVD_MAL_CASE	69	79	59	21	37	11	CVD_MAL_CTRL	34	43	25	9	25	0
1988	C	T	HELD_ALL_BAD	100	143	57	52	39	9	HELD_ALL_GOOD	116	144	88	48	48	20
2000	C	T	CVD_MAL_CASE	70	136	4	68	2	0	CVD_MAL_CTRL	34	58	10	29	5	0
2000	C	T	CVD_ALL_CASE	105	202	8	101	4	0	CVD_ALL_CTRL	74	130	18	65	9	0
2000	C	T	HELD_FEM_CASE2	46	90	2	45	1	0	HELD_FEM_CTRL2	42	74	10	37	5	0
2000	C	T	HELD_MAL_LOHDL	20	40	0	20	0	0	HELD_MAL_HHDL	22	40	4	20	2	0
2000	C	T	HELD_FEM_ADRCASE	79	154	4	77	2	0	HELD_FEM_ADRCTRL	82	152	12	76	6	0
2000	C	T	HELD_MAL_CASE	14	22	6	11	3	0	HELD_MAL_CTRL	19	36	2	18	1	0
2071	A	G	CVD_ALL_CASE	102	80	124	14	52	36	CVD_ALL_CTRL	74	42	106	4	34	36
2078	G	T	HELD_MAL_BAD	18	13	23	1	11	6	HELD_MAL_GOOD	35	13	57	0	13	22
2085	G	T	HELD_FEM_VHRESP	10	16	4	6	4	0	HELD_FEM_VLORESP	14	13	15	3	7	4
2095	A	G	CVD_ALL_CASE	105	4	206	4	101	0	CVD_ALL_CTRL	73	146	0	0	73	0
2119	A	G	HELD_MAL_BAD	20	23	17	3	17	0	HELD_MAL_GOOD	37	53	21	16	21	0
2119	A	G	HELD_ALL_BAD	102	131	73	29	73	0	HELD_ALL_GOOD	117	166	68	49	68	0
2119	A	G	HELD_FEM_HIRESP	12	15	9	3	9	0	HELD_FEM_LORESP	22	35	9	13	9	0
2141	A	G	HELD_FEM_VHRESP	12	6	18	0	6	6	HELD_FEM_LORESP	22	6	38	2	2	18
2141	A	G	HELD_ALL_CASE	45	17	73	0	17	28	HELD_ALL_CTRL	39	15	63	3	9	27
2182	A	G	HELD_FEM_HIRESP	12	18	6	6	6	0	HELD_FEM_LORESP	21	16	26	1	14	6
2234	G	T	HELD_MAL_BAD	20	10	30	0	10	10	HELD_MAL_GOOD	35	32	38	7	18	10
2281	A	C	HELD_FEM_VHRESP	9	5	13	0	5	4	HELD_FEM_VLORESP	13	15	11	4	7	2
2298	A	C	CVD_FEM_CASE	35	18	52	4	10	21	CVD_FEM_CTRL	38	20	56	0	20	18
2298	A	C	HELD_MAL_CASE2	29	8	50	0	8	21	HELD_MAL_CTRL2	28	16	40	2	12	14

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baySNP	A1	A2	COHORTA	SIZE	FQ1_A	FQ2_A	FQ11	FQ12	FQ22	COHORT_B	SIZE	FQ1_B	FQ2_B	FQ11	FQ12	FQ22
	A	A	A	A	A	A	A	A	A	B	B	B	B	B	B	B
2341	C	T	HELD_FEM_CASE	31	6	56	0	6	25	HELD_FEM_CTRL	22	44	0	0	0	22
2357	A	G	HELD_ALL_CASE2	74	28	120	5	18	51	HELD_ALL_CTRL2	71	25	117	0	25	46
2357	A	G	HELD_ALL_CASE	45	16	74	4	8	33	HELD_ALL_CTRL	40	14	66	0	14	26
2357	A	G	HELD_MAL_BAD	20	4	36	0	4	16	HELD_MAL_GOOD	36	17	55	0	17	19
2357	A	G	HELD_FEM_CASE	31	12	50	4	4	23	HELD_FEM_CTRL	22	7	37	0	7	15
2366	G	T	CVD_FEM_CASE	33	38	28	12	14	7	CVD_FEM_CTRL	40	31	49	8	15	17
2423	A	G	CVD_FEM_CASE	33	45	21	16	13	4	CVD_FEM_CTRL	39	38	40	12	14	13
2708	C	T	CVD_FEM_CASE	29	57	1	28	1	0	CVD_FEM_CTRL	40	73	7	33	7	0
2995	A	C	HELD_FEM_AdRCASE5	18	16	20	3	10	5	HELD_FEM_AdRCRTL	82	45	119	4	37	41
			ULN													
2995	A	C	HELD_FEM_UHIRESP	54	24	84	2	20	32	HELD_FEM_ULORESP	75	50	100	5	40	30
3360	G	T	HELD_MAL_AdRCASE5	10	20	0	10	0	0	HELD_MAL_AdRCRTL	73	122	24	50	22	1
			ULN													
3464	A	G	HELD_ALL_CASE	45	21	69	3	15	27	HELD_ALL_CTRL	40	35	45	9	17	14
3464	A	G	HELD_FEM_CASE	31	13	49	3	7	21	HELD_FEM_CTRL	22	19	25	5	9	8
3689	C	G	HELD_FEM_HIRESP	6	9	3	3	0	0	HELD_FEM_LORESP	14	10	18	1	8	5
3975	A	C	HELD_FEM_UHIRESP	56	28	84	2	24	30	HELD_FEM_ULORESP	75	58	92	10	38	27
3976	A	G	HELD_FEM_UHIRESP	56	28	84	2	24	30	HELD_FEM_ULORESP	75	57	93	11	35	29
4206	A	T	HELD_FEM_AdRCASE3	37	36	38	8	20	9	HELD_FEM_AdRCRTL	83	103	63	31	41	11
			ULN													
4838	A	G	HELD_FEM_VHIRESP	10	16	4	7	2	1	HELD_FEM_VLORESP	14	14	14	3	8	3
4838	A	G	HELD_FEM_VHIRESP	10	16	4	7	2	1	HELD_FEM_VLORESP	14	14	14	3	8	3

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KeySNP	A1	A2	COHORTA	SIZE	FQ1A	FQ2A	FQ11	FQ12	FQ22	COHORTB	SIZE	FQ1B	FQ2B	FQ11	FQ12	FQ22
5717	A	G	HELD_ALL_ADRCASE3	65	74	56	21	32	12	HELD_ALL_ADRCTRL	156	144	168	34	76	46
5959	A	G	HELD_ALL_CASE	43	52	34	16	20	7	HELD_ALL_CTRL	38	29	47	4	21	13
5959	A	G	CVD_FEM_CASE	9	12	6	4	4	1	CVD_FEM_CTRL	13	7	19	0	7	6
5959	A	G	HELD_MAL_CASE	14	15	13	4	7	3	HELD_MAL_CTRL	17	10	24	0	10	7
5959	A	G	HELD_MAL_ADRCASE5	9	6	12	2	2	5	HELD_MAL_ADRCTRL	67	67	13	41	13	
5959	A	G	HELD_FEM_ADRCASE	72	71	73	15	41	16	HELD_FEM_ADRCTRL	68	51	85	11	29	28
6162	C	G	HELD_ALL_ADRCASE3	64	37	91	1	35	28	HELD_ALL_ADRCTRL	151	90	212	19	52	80
6162	C	G	HELD_ALL_ADRCASE	156	88	224	6	76	74	HELD_ALL_ADRCTRL	151	90	212	19	52	80
6162	C	G	HELD_ALL_ADRCASE5	27	16	38	0	16	11	HELD_ALL_ADRCTRL	151	90	212	19	52	80
6162	C	G	HELD_MAL_ADRCASE3	26	13	39	0	13	13	HELD_MAL_ADRCTRL	71	43	99	11	21	39
6162	C	G	HELD_FEM_ADRCASE5	18	13	23	0	13	5	HELD_FEM_ADRCTRL	80	47	113	8	31	41
6162	C	G	HELD_MAL_ADRCASE	74	40	108	3	34	37	HELD_MAL_ADRCTRL	71	43	99	11	21	39
6236	C	T	HELD_ALL_ADRCASE5	27	24	30	6	12	9	HELD_ALL_ADRCTRL	152	84	220	13	58	81
6236	C	T	HELD_MAL_ADRCASE3	27	23	31	4	15	8	HELD_MAL_ADRCTRL	72	38	106	5	28	39

baySNP	A1	A2	COHORT_A	SIZE	FQ1_A	FQ2_A	FQ11	FQ12	FQ22	COHORT_B	SIZE	FQ1_B	FQ2_B	FQ11	FQ12	FQ22
	A	A	A	A	A	A	A	A	A	B	B	B	B	B	B	B
6236	C	T	HELD_MAL_ADRCASE3	10	10	10	2	6	2	HELD_MAL_ADRCTRL	72	38	106	5	28	39
			ULN													
6236	C	T	HELD_ALL_ADRCASE3	63	47	79	10	27	26	HELD_ALL_ADRCTRL	152	84	220	13	58	81
			ULN													
6482	A	G	HELD_MAL_LOHDL	17	18	16	5	8	4	HELD_MAL_HHDL	21	34	8	15	4	2
6482	A	G	HELD_ALL_BAD2	619	918	320	238	41	41	HELD_ALL_GOOD2	709	1098	320	436	226	47
6482	A	G	HELD_MAL_CASE2	27	43	11	18	7	2	HELD_MAL_CTRL2	28	32	24	10	12	6
6482	A	G	HELD_MAL_BAD2	309	461	157	173	21	21	HELD_MAL_GOOD2	339	539	139	220	99	20
6498	A	G	CVD_FEM_CASE	32	60	4	28	4	0	CVD_FEM_CTRL	35	57	13	25	7	3
6744	C	T	HELD_ALL_ADRCASE	26	21	31	4	13	9	HELD_ALL_ADRCTRL	149	74	224	9	56	84
			ULN													
7133	C	G	HELD_MAL_CASE	14	20	8	10	0	4	HELD_MAL_CTRL	18	36	0	18	0	0
8021	A	G	CVD_FEM_CASE	28	35	21	8	19	1	CVD_FEM_CTRL	36	44	28	15	14	7
8060	A	G	CVD_FEM_CASE	35	65	5	31	3	1	CVD_FEM_CTRL	40	68	12	28	12	0
8060	A	G	HELD_FEM_LOHDL	18	29	7	11	7	0	HELD_FEM_HHDL	23	43	3	20	3	0
8210	A	G	HELD_FEM_HIRESP	12	9	15	1	7	4	HELD_FEM_LORESP	22	22	22	9	4	9
8592	C	T	HELD_FEM_VHIRESP	150	122	178	15	92	43	HELD_FEM_VLORESP	143	118	168	25	68	50
8816	C	G	HELD_FEM_HIRESP	13	15	11	4	7	2	HELD_FEM_LORESP	11	5	17	0	5	6
8846	A	G	HELD_ALL_BAD	107	161	53	57	47	3	HELD_ALL_GOOD	116	166	66	62	42	12
8943	A	C	HELD_FEM_BAD	20	35	5	15	5	0	HELD_MAL_GOOD	37	52	22	20	12	5
9193	C	G	CVD_FEM_CASE	83	155	11	72	11	0	HELD_FEM_GOOD	80	140	20	60	20	0
9193	C	G	CVD_FEM_CTRL	36	63	9	28	7	1	CVD_FEM_CTRL	40	77	3	37	3	0

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DaySNP	A1	A2	COHORT A	SIZE	FQ1A	FQ2A	FQ11	FQ12	FQ22	COHORT B	SIZE	FQ1B	FQ2B	FQ11	FQ12	FQ22
	A	A	A	A	A	A	A	A	A	B	B	B	B	B	B	B
9443	C	T	CVD_MAL_CASE	69	43	95	9	25	35	CVD_MAL_CTRL	33	12	54	0	12	21
9516	A	G	HELD_MAL_CASE	14	17	11	7	3	4	HELD_MAL_CTRL	18	12	24	2	8	8
9698	A	G	HELD_MAL_ADRCASE	74	8	140	4	0	70	HELD_MAL_ADRCTRL	72	30	114	14	2	56
9698	A	G	HELD_MAL_ADRCASE3	27	54	0	0	0	27	HELD_MAL_ADRCTRL	72	30	114	14	2	56
			ULN													
9698	A	G	HELD_FEM_HIRESP	294	105	483	5	95	194	HELD_FEM_LORESP	298	123	473	16	91	191
9698	A	G	HELD_MAL_ADRCASE5	10	20	0	0	0	10	HELD_MAL_ADRCTRL	72	30	114	14	2	56
			ULN													
9698	A	G	CVD_ALL_CASE	102	46	158	17	12	73	CVD_ALL_CTRL	72	19	125	6	7	59
9849	C	T	HELD_FEM_CASE	31	62	0	31	0	0	HELD_FEM_CTRL	21	39	3	18	3	0
9849	C	T	HELD_MAL_BAD	20	35	5	15	5	0	HELD_MAL_GOOD	37	72	2	35	2	0
9883	A	G	HELD_FEM_CASE	31	23	39	7	9	15	HELD_FEM_CTRL	22	18	26	1	16	5
9883	A	G	HELD_ALL_CASE	45	33	57	9	15	21	HELD_ALL_CTRL	39	32	46	4	24	11
10079	A	G	CVD_ALL_CASE	103	8	198	4	0	99	CVD_ALL_CTRL	73	1	145	0	1	72
10079	A	G	CVD_MAL_CASE	68	8	128	4	0	64	CVD_MAL_CTRL	34	68	0	0	0	34
10481	A	T	HELD_FEM_ADRCASE5	17	12	22	3	6	8	HELD_FEM_ADRCTRL	83	97	69	32	33	18
			ULN													
10542	C	T	HELD_FEM_UHIRESP	54	8	100	1	6	47	HELD_FEM_ULORESP	75	21	129	0	21	54
10542	C	T	HELD_MAL_ADRCASE5	10	20	0	0	0	10	HELD_MAL_ADRCTRL	69	14	124	0	14	55
			ULN													
10600	A	G	HELD_FEM_HIRESP	21	42	0	0	0	21	HELD_FEM_LORESP	33	4	62	0	4	29
10621	C	T	HELD_FEM_CASE	30	52	8	24	4	2	HELD_FEM_CTRL	20	32	8	12	8	0

baySNP	A1	A2	COHORT A	SIZE	FQ1A	FQ2A	FQ11	FQ12	COHORT B	SIZE	FQ1B	FQ2B	FQ11	FQ12	EQ22	
	A	A	A	A	A	A	A	A	B	B	B	B	B	B	B	
10745	A	G	HELD_ALL_ADRCASE5	27	20	34	5	10	12	HELD_ALL_ADRCTRL	148	75	221	7	61	80
			ULN													
10745	A	G	HELD_FEM_VHRESP	153	90	216	11	68	74	HELD_FEM_VLORESP	150	77	223	16	45	89
10747	C	T	HELD_MAL_ADRCASE	76	74	78	14	46	16	HELD_MAL_ADRCTRL	70	64	76	3	58	9
10747	C	T	CVD_ALL_CASE	62	54	70	15	24	23	CVD_ALL_CTRL	74	51	97	6	39	29
10747	C	T	HELD_MAL_ADRCASE3	27	24	30	4	16	7	HELD_MAL_ADRCTRL	70	64	76	3	58	9
			ULN													
10771	C	G	HELD_MAL_ADRCASE5	10	12	8	4	4	2	HELD_MAL_ADRCTRL	70	48	92	6	36	28
			ULN													
10771	C	G	HELD_FEM_HIRESP	284	222	346	52	118	114	HELD_FEM_LORESP	276	185	367	40	105	131
10870	A	G	HELD_MAL_BAD	20	11	29	0	11	9	HELD_MAL_GOOD	37	19	55	5	9	23
10870	A	G	HELD_FEM_BAD	82	32	132	7	18	57	HELD_FEM_GOOD	77	46	108	8	30	39
10870	A	G	HELD_MAL_CASE	14	3	25	0	3	11	HELD_MAL_CTRL	18	12	24	2	8	8
10870	A	G	HELD_ALL_CASE	45	17	73	2	13	30	HELD_ALL_CTRL	40	27	53	6	15	19
10877	A	C	HELD_ALL_LOHDL	9	18	0	0	0	9	HELD_ALL_HIHDL	15	7	23	1	5	9
10948	G	T	HELD_FEM_BAD	84	83	85	16	51	17	HELD_FEM_GOOD	79	95	63	31	33	15
10948	G	T	HELD_ALL_BAD	104	104	104	22	60	22	HELD_ALL_GOOD	115	138	92	44	50	21
10948	G	T	HELD_FEM_CASE2	44	46	42	9	28	7	HELD_FEM_CTRL2	42	50	34	17	16	9
10948	G	T	CVD_MAL_CASE	69	63	75	12	39	18	CVD_MAL_CTRL	34	41	27	12	17	5
11001	C	T	HELD_MAL_ADRCASE5	10	9	11	2	5	3	HELD_MAL_ADRCTRL	75	41	109	2	37	36
			ULN													
11073	C	G	HELD_MAL_ADRCASE5	9	10	8	3	4	2	HELD_MAL_ADRCTRL	68	43	93	9	25	34

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hapSNP	A1	A2	COHORT_A	SIZE	FQ1_A	FQ2_A	FQ11	FQ12	FQ22	COHORT_B	SIZE	FQ1_B	FQ2_B	FQ11	FQ12	FQ22
			A	A	A	A	A	A	A	B	B	B	B	B	B	B
			ULN													
11153	C	T	HELD_FEM_CASE	31	55	7	24	7	0	HELD_FEM_CTRL	22	33	11	11	11	0
11210	C	T	HELD_MAL_CASE	14	23	5	9	5	0	HELD_MAL_CTRL	19	37	1	18	1	0
11210	C	T	HELD_ALL_ADRCASE3	63	110	16	47	16	0	HELD_ALL_ADRCTRL	144	267	21	125	17	2
			ULN													
11210	C	T	HELD_ALL_ADRCASE	153	275	31	122	31	0	HELD_ALL_ADRCTRL	144	267	21	125	17	2
11248	C	T	HELD_FEM_ADRCASE	81	131	31	56	19	6	HELD_FEM_ADRCTRL	79	112	46	38	36	5
11248	C	T	HELD_MAL_BAD	18	33	3	15	3	0	HELD_MAL_GOOD	34	53	15	19	15	0
11248	C	T	HELD_ALL_CASE	41	68	14	27	14	0	HELD_ALL_CTRL	31	44	18	13	18	0
11372	A	G	HELD_MAL_BAD	20	25	15	10	5	5	HELD_MAL_GOOD	36	31	41	10	11	15
11449	C	G	HELD_FEM_CASE	31	6	56	1	4	26	HELD_FEM_CTRL	22	10	34	0	10	12
11450	A	T	HELD_FEM_HRESP	289	170	408	28	114	147	HELD_FEM_LORESP	290	139	441	16	107	167
11470	C	T	HELD_MAL_BAD	20	40	0	20	0	0	HELD_MAL_GOOD	36	67	5	31	5	0
11472	A	T	HELD_MAL_BAD	20	40	0	20	0	0	HELD_MAL_GOOD	35	65	5	30	5	0
11472	A	T	HELD_FEM_BAD	83	158	8	75	8	0	HELD_FEM_GOOD	80	158	2	78	2	0
11487	A	T	HELD_MAL_ADRCASE5	10	20	0	0	10	0	HELD_MAL_CTRL	69	34	104	34	35	0
			ULN													
11487	A	T	HELD_MAL_ADRCASE3	27	6	48	6	21	0	HELD_MAL_CTRL	69	34	104	34	35	0
			ULN													
11488	C	G	HELD_MAL_ADRCASE5	10	20	0	10	0	0	HELD_MAL_CTRL	70	102	38	35	32	3
			ULN													
11488	C	G	HELD_FEM_UHRESP	54	78	30	29	20	5	HELD_FEM_ULORESP	77	126	28	49	28	0

baySNP	A1	A2	COHORT_A	SIZE	FQ1_A	FQ2_A	FQ11	FQ12	FQ22	COHORT_B	SIZE	FQ1_B	FQ2_B	FQ11	FQ12	FQ22
	A	A	A	A	A	A	A	A	A	B	B	B	B	B	B	B
11488	C	G	HELD_MAL_ADRCASE3	26	44	8	20	4	2	HELD_MAL_ADRCTRL	70	102	38	35	32	3
11493	A	G	HELD_MAL_CASE	14	6	22	0	6	8	HELD_MAL_CTRL	18	6	30	2	2	14
11502	C	T	HELD_MAL_ADRCASE3	27	8	46	0	8	19	HELD_MAL_ADRCTRL	73	44	102	7	30	36
11502	C	T	HELD_MAL_ADRCASE5	10	2	18	0	2	8	HELD_MAL_ADRCTRL	73	44	102	7	30	36
11534	G	T	HELD_ALL_BAD	102	204	0	102	0	0	HELD_ALL_GOOD	117	231	3	114	3	0
11537	A	G	CVD_FEM_CASE	36	52	20	20	12	4	CVD_FEM_CTRL	39	68	10	30	8	1
11537	A	G	HELD_FEM_HIRESP	12	22	2	10	2	0	HELD_FEM_LORESP	22	31	13	12	7	3
11560	A	G	HELD_FEM_HIRESP	12	2	22	1	0	11	HELD_FEM_LORESP	22	44	0	0	0	22
11578	C	T	HELD_FEM_BAD	61	121	1	60	1	0	HELD_FEM_GOOD	65	122	8	57	8	0
11578	C	T	CVD_FEM_CASE	30	57	3	27	3	0	CVD_FEM_CTRL	39	78	0	39	0	0
11594	C	T	HELD_FEM_ADRCASE3	37	74	0	0	0	37	HELD_FEM_ADRCTRL	80	10	150	2	6	72
11594	C	T	HELD_ALL_ADRCASE5	27	54	0	0	0	27	HELD_ALL_ADRCTRL	151	20	282	2	16	133
11594	C	T	HELD_ALL_CASE	45	10	80	0	10	35	HELD_ALL_CTRL	41	3	79	0	3	38
11594	C	T	HELD_ALL_ADRCASE	155	9	301	1	7	147	HELD_ALL_ADRCTRL	151	20	282	2	16	133
11594	C	T	HELD_FEM_ADRCASE5	18	36	0	0	0	18	HELD_FEM_ADRCTRL	80	10	150	2	6	72
11624	C	T	HELD_ALL_CASE	42	57	27	21	15	6	HELD_ALL_CTRL	40	60	20	20	20	0

baySNP	A1	A2	COHORT A	SIZE	FQ1 A		FQ2 A		FQ11 A		FQ12 A		COHORT B		FQ1 B		FQ2 B		FQ11 B		FQ12 B	
					A	A	A	A	A	A	A	A	B	B	B	B	B	B	B	B	B	
11656	C	T	HELD_MAL_BAD	20	20	6	8	6	6	HELD_MAL_GOOD	36	53	19	19	15	15	2	2	2	2	2	
11656	C	T	HELD_FEM_HIRESP	12	19	5	7	5	0	HELD_FEM_LORESP	22	24	20	5	14	14	3	3	3	3	3	
11656	C	T	HELD_ALL_BAD	102	119	85	35	49	18	HELD_ALL_GOOD	114	156	72	51	54	54	9	9	9	9	9	
11825	A	G	HELD_MAL_DRCCASE5	9	15	3	6	3	0	HELD_MAL_DRCTRL	63	121	5	58	5	58	0	0	0	0	0	
11914	A	T	HELD_MAL_DRCCASE5	9	2	16	1	0	8	HELD_MAL_DRCTRL	69	83	55	41	1	41	1	27	27	27	27	
11914	A	T	HELD_ALL_DRCCASE5	27	24	30	6	12	9	HELD_ALL_DRCTRL	151	178	124	63	52	52	36	36	36	36	36	
12008	C	T	HELD_FEM_HIRESP	278	529	27	251	27	0	HELD_FEM_LORESP	277	541	13	264	13	264	0	0	0	0	0	
12008	C	T	HELD_ALL_DRCCASE5	24	48	0	24	0	0	HELD_ALL_DRCTRL	134	256	12	122	12	122	0	0	0	0	0	
12097	A	G	HELD_ALL_DRCCASE5	28	6	50	6	22	0	HELD_ALL_DRCTRL	155	11	299	11	144	11	144	0	0	0	0	
12097	A	G	HELD_FEM_DRCCASE3	38	7	69	7	31	0	HELD_FEM_DRCTRL	83	5	161	5	78	5	78	0	0	0	0	
12097	A	G	HELD_MAL_DRCCASE5	10	3	17	3	7	0	HELD_MAL_DRCTRL	72	6	138	6	66	6	66	0	0	0	0	
12097	A	G	HELD_ALL_DRCCASE3	63	10	116	10	53	0	HELD_ALL_DRCTRL	155	11	299	11	144	11	144	0	0	0	0	
12366	A	G	HELD_FEM_UHRESP	50	82	18	32	18	0	HELD_FEM_ULORESP	74	104	44	39	26	39	26	9	9	9	9	
12366	A	G	HELD_ALL_DRCCASE5	25	40	10	18	4	3	HELD_ALL_DRCTRL	151	229	73	85	59	85	59	7	7	7	7	

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BaySNP	A1	A2	COHORT A	SIZE	FQ1_A	FQ2_A	FQ11	FQ12	FQ22	COHORT B	SIZE	FQ1_B	FQ2_B	FQ11	FQ12	FQ22
	A	A	ULN		A	A	A	A				B	B	B	B	B
12619	A	G	HELD_MAL_ADRCASE5	10	19	1	9	0		HELD_MAL_ADRCTRL	71	142	0	0	71	0
12619	A	G	HELD_ALL_ADRCASE5	27	52	2	25	0		HELD_ALL_ADRCTRL	151	1	301	1	150	0
13025	A	C	HELD_ALL_ADRCASE5	28	34	22	13	8	7	HELD_ALL_ADRCTRL	151	201	101	65	71	15
13191	A	G	HELD_FEM_BAD	83	42	124	6	30	47	HELD_FEM_GOOD	79	62	96	10	42	27
13191	A	G	HELD_MAL_CASE	14	11	17	2	7	5	HELD_MAL_CTRL	18	5	31	0	5	13
13191	A	G	HELD_ALL_BAD	101	51	151	6	39	56	HELD_ALL_GOOD	114	81	147	13	55	46
13937	A	C	HELD_FEM_ADRCASE5	17	19	15	4	11	2	HELD_FEM_ADRCTRL	83	122	44	42	38	3
			ULN													
900002	G	T	CVD_FEM_CASE	34	23	45	5	13	16	CVD_FEM_CTRL	40	15	65	2	11	27
900013	C	G	CVD_FEM_CASE	35	49	21	20	9	6	CVD_FEM_CTRL	40	49	31	13	23	4
900013	C	G	CVD_ALL_CASE	104	150	58	58	34	12	CVD_ALL_CTRL	74	97	51	29	39	6
900025	G	T	CVD_MAL_CASE	66	41	91	7	27	32	CVD_MAL_CTRL	34	31	37	7	17	10
900032	C	T	CVD_FEM_CASE	25	47	3	23	1	1	CVD_FEM_CTRL	37	65	9	28	9	0
900045	C	T	HELD_FEM_HIRESP	12	4	20	1	2	9	HELD_FEM_LORESP	22	18	26	5	8	9
900065	A	C	CVD_FEM_CASE	32	54	10	22	10	0	CVD_FEM_CTRL	39	50	28	16	18	5
900065	A	C	CVD_MAL_CASE	59	80	38	25	30	4	CVD_MAL_CTRL	29	36	22	7	22	0
900065	A	C	CVD_ALL_CASE	91	134	48	47	40	4	CVD_ALL_CTRL	68	86	50	23	40	5
900078	A	G	HELD_ALL_ADRCASE3	64	116	12	52	12	0	HELD_ALL_ADRCTRL	155	297	13	142	13	0

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laysNP	A1	A2	COHORT A	SIZE A	FQ1A	FQ2A	FQ12	FQ22	COHORT B	SIZE B	FQ1B	FQ2B	FQ11	FQ21	FQ12	FQ22
			ULN													
900078	A	G	HELD_ALL_ADRCASES	27	48	6	21	6	0	HELD_ALL_ADRCTRL	155	297	13	142	13	0
900078	A	G	HELD_FEM_ADRCASE3	38	69	7	31	7	0	HELD_FEM_ADRCTRL	83	161	5	78	5	0
900082	A	G	HELD_FEM_ADRCASE3	35	25	45	8	9	18	HELD_FEM_ADRCTRL	74	70	78	17	36	21
900082	A	G	HELD_FEM_ADRCASE5	17	10	24	3	4	10	HELD_FEM_ADRCTRL	74	70	78	17	36	21
900096	A	G	CVD_ALL_CASE	101	157	45	60	37	4	CVD_ALL_CTRL	72	125	19	55	15	2
900107	C	T	HELD_MAL_ADRCASE5	10	2	18	0	2	8	HELD_MAL_ADRCTRL	73	43	103	9	25	39
900115	A	G	HELD_MAL_ADRCASE5	9	6	12	1	4	4	HELD_MAL_ADRCTRL	72	91	53	27	37	8
900115	A	G	HELD_FEM_HIRESP	40	58	22	22	14	4	HELD_FEM_LORESP	46	62	30	17	28	1
900121	G	T	HELD_MAL_ADRCASE	66	47	85	5	37	24	HELD_MAL_ADRCTRL	67	56	78	15	26	26
900173	G	T	CVD_ALL_CASE	23	17	29	5	7	11	CVD_ALL_CTRL	22	26	18	11	4	7
10000002	A	G	HELD_FEM_HIRESP	12	21	3	9	3	0	HELD_FEM_LORESP	22	25	19	9	7	6
10000006	A	G	HELD_FEM_CASE	31	58	4	28	2	1	HELD_FEM_CTRL	22	31	13	11	9	2

baysNP	A1	A2	COHORTA	SIZE	FQ1A	FQ2A	FQ11	FQ12	FQ22	COHORTB	SIZE	FQ1B	FQ2B	FQ11	FQ12	FQ22
	A	A		A	A	A	A	A	A		B	B	B	B	B	B
1000006	A	G	HELD_ALL_CASE	44	82	6	39	4	1	HELD_ALL_CTRL	38	58	18	23	12	3
1000014	A	C	HELD_ALL_CASE	45	83	7	40	3	2	HELD_ALL_CTRL	39	64	14	26	12	1
1000014	A	C	HELD_FEM_CASE	31	58	4	28	2	1	HELD_FEM_CTRL	22	37	7	15	7	0
1000025	C	T	HELD_MAL_BAD	20	29	11	9	11	0	HELD_MAL_GOOD	36	43	29	14	15	7

Table 5b p-values of PA SNPs. A SNP is considered as associated to cardiovascular disease, adverse statin response or to efficacy of statin treatment, respectively, when one of the p values is equal or below 0.05.

BAYSNP	COMPARISON	GTYPE CPVAL	GTYPE LRPVAL	GTYPE XPVAL	ALLELE CPVAL	ALLELE XPVAL	ALLELE LRPVAL
28	HELD_FEM_EFF	0,0506	0,0508	0,0442	0,0411	0,0579	0,0349
29	HELD_ALL_HDL	0,021	0,0227	0,0099	0,0089	0,0164	0,0087
29	HELD_MAL_ADDR3ULN	0,0602	0,0582	0,0664	0,0446	0,0526	0,0435
29	HELD_MAL_ADDR5ULN	0,1406	0,1835	0,1554	0,0455	0,0778	0,0422
52	HELD_FEM_EFF	0,0644	0,0861	0,0488	0,0272	0,0362	0,0261
56	HELD_FEM_EFF	0,0248	0,0379	0,0273	0,0347	0,0479	0,0393
89	HELD_ALL_CC	0,0614	0,1	0,0311	0,0638	0,1021	0,0323
90	HELD_FEM_CC	0,0398	0,0424	0,0242	0,1382	0,1687	0,137
99	HELD_FEM_LP	0,0363	0,0366	0,0338	0,8397	0,9056	0,8397
140	HELD_FEM_EFF	0,3895	0,6921	0,2368	0,1188	0,288	0,0524
152	HELD_FEM_EFF	0,1084	0,1216	0,1082	0,0373	0,0595	0,0389
214	HELD_ALL_LP	0,1139	0,1152	0,0532	0,9756	1	0,9756

BAYSNP	COMPARISON	GTYPE CPVAL	GTYPE XPVAL	GTYPE LRPVAL	ALLELE CPVAL	ALLELE XPVAL	ALLELE LRPVAL
214	HELD_FEM_LIP	0,1095	0,1196	0,0506	0,5567	0,5803	0,5567
221	HELD_ALL_CC	0,0367	0,0359	0,0353	0,4257	0,506	0,426
221	HELD_FEM_CC	0,0406	0,0424	0,0384	0,1456	0,2083	0,1469
224	HELD_FEM_LIP	0,2893	0,3016	0,2874	0,0533	0,0709	0,0527
224	HELD_MAL_LIP	0,2292	0,2815	0,1975	0,0278	0,0392	0,0221
294	HELD_ALL_CC	0,0851	0,1041	0,0327	0,1547	0,1913	0,1534
307	CVD_FEM	0,013	0,0118	0,0104	0,0032	0,004	0,003
307	HELD_ALL_LIP	0,0255	0,0273	0,0249	0,0934	0,0968	0,0936
411	HELD_ALL_HDL	0,1529	0,2195	0,1076	0,0588	0,1136	0,0513
449	HELD_MAL_LIP	0,1321	0,0942	0,1001	0,0535	0,0667	0,0416
466	CVD_FEM	0,133	0,1439	0,1301	0,0444	0,0505	0,0438
472	HELD_FEM_EFF	0,0453	0,0626	0,0116	0,0068	0,0146	0,0009
542	HELD_MAL_CC	0,0014	0,0009	0,0007	0,0002	0,0003	0,0002
542	HELD_MAL_HDL	0,0054	0,0028	0,0029	0,0004	0,0005	0,0003
542	HELD_ALL_ADR	0,0257	0,0152	0,0171	0,0971	0,1108	0,0962
542	HELD_FEM_HDL	0,1914	0,1661	0,1457	0,0613	0,0709	0,0487
739	HELD_ALL_CC	0,0958	0,0983	0,0902	0,03	0,0327	0,0296
821	HELD_MAL_LIP2	0,0426	0,0436	0,0419	0,0865	0,0927	0,0867
821	HELD_FEM_VEFF	0,1193	0,1222	0,0584	0,0343	0,0681	0,0306
1005	HELD_MAL_CC	0,2376	0,3423	0,1618	0,0603	0,0946	0,0502
1055	HELD_MAL_CC	0,0302	0,0328	0,0084	0,2241	0,2988	0,216

BAYSNP	COMPARISON	GTYPE CPVAL	GTYPE XPVAL	GTYPE LRPVAL	ALLELE CPVAL	ALLELE XPVAL	ALLELE LRPVAL
1056	HELD_FEM_EFF	0,0094	0,0085	0,0079	0,9671	1	0,9671
1085	HELD_MAL_LIP	0,0889	0,0964	0,0773	0,0288	0,0462	0,0288
1085	CVD_FEM	0,1655	0,1833	0,156	0,0373	0,0546	0,0359
1086	HELD_MAL_LIP	0,0963	0,1125	0,0928	0,0318	0,0475	0,0315
1092	HELD_MAL_LIP	0,0493	0,0492	0,046	0,0712	0,0958	0,0663
1096	HELD_MAL_CC	0,0436	0,0623	0,0423	0,0685	0,0895	0,0679
1096	CVD_MAL	0,0766	0,0645	0,0452	0,5906	0,6848	0,5926
1101	HELD_FEM_EFF	0,1158	0,2728	0,0522	0,1279	0,2891	0,0572
1204	HELD_MAL_LIP	0,0471	0,0447	0,0362	0,0189	0,0238	0,0214
1204	HELD_ALL_LIP	0,1563	0,1592	0,1558	0,0422	0,0485	0,0424
1504	HELD_ALL_CC	0,0128	0,0133	0,0115	0,5946	0,64	0,5946
1504	HELD_MAL_LIP	0,0864	0,087	0,0247	0,1834	0,2241	0,1799
1504	HELD_MAL_CC	0,051	0,0757	0,0467	0,2868	0,3134	0,2871
1504	HELD_FEM_CC	0,0535	0,0663	0,0532	0,0878	0,1084	0,0873
1511	HELD_FEM_EFF	0,0513	0,0299	0,0413	0,1279	0,1563	0,1329
1524	HELD_FEM_DR3ULN	0,0684	0,0673	0,0215	0,64	0,7419	0,6382
1556	HELD_FEM_EFF	0,0063	0,0151	0,0066	0,0129	0,0269	0,015
1561	CVD_FEM	0,1299	0,1484	0,1216	0,0472	0,0666	0,0456
1582	HELD_MAL_LIP	0,1444	0,1408	0,0649	0,0389	0,0633	0,0319
1638	HELD_FEM_CC	0,0876	0,0903	0,0861	0,0318	0,0385	0,0328
1653	CVD_MAL	0,0269	0,0234	0,0255	0,4812	0,5499	0,4809

BAYSNP	COMPARISON	GTYPE CPVAL	GTYPE XPVAL	GTYPE LRPVAL	ALLELE CPVAL	ALLELE XPVAL	ALLELE LRPVAL
1662	HELD_MAL_CC	0,0153	0,0278	0,0067	0,0006	0,0007	0,0001
1714	CVD_MAL	0,0716	0,0776	0,0817	0,0388	0,0484	0,041
1722	HELD_FEM_ADR5ULN	0,0325	0,0304	0,0429	0,1144	0,1401	0,1146
1757	HELD_FEM_EFF	0,0289	0,0296	0,0153	0,1752	0,1926	0,1779
1765	HELD_ALL_ADR3ULN	0,0044	0,0049	0,0024	0,0023	0,0029	0,0012
1765	HELD_ALL_ADR3ULN	0,0044	0,0049	0,0024	0,0023	0,0029	0,0012
1765	HELD_ALL_ADR5ULN	0,0469	0,0457	0,0235	0,0166	0,0163	0,0077
1765	HELD_ALL_ADR5ULN	0,0469	0,0457	0,0235	0,0166	0,0163	0,0077
1765	HELD_MAL_ADR3ULN	0,0428	0,0505	0,0211	0,0131	0,0174	0,0058
1765	HELD_MAL_ADR3ULN	0,0428	0,0505	0,0211	0,0131	0,0174	0,0058
1765	HELD_MAL_ADR5ULN	0,0997	0,0786	0,0255	0,0396	0,0451	0,0069
1765	HELD_MAL_ADR5ULN	0,0997	0,0786	0,0255	0,0396	0,0451	0,0069
1765	HELD_FEM_ADR3ULN	0,0666	0,0733	0,0522	0,0513	0,0579	0,0423
1765	HELD_FEM_ADR3ULN	0,0666	0,0733	0,0522	0,0513	0,0579	0,0423
1776	HELD_ALL_CC	0,0614	0,1	0,0311	0,0082	0,0098	0,0023
1776	HELD_FEM_CC	0,087	0,1676	0,0568	0,0155	0,0273	0,0071
1799	HELD_FEM_LIP2	0,006	0,0058	0,0061	0,2598	0,268	0,2601
1799	HELD_MAL_CC	0,1419	0,1545	0,134	0,0408	0,0604	0,0406
1806	HELD_FEM_EFF	0,1946	0,236	0,128	0,047	0,0817	0,0299
1837	HELD_FEM_LIP2	0,0049	0,0047	0,0048	0,569	0,5843	0,5688
1837	HELD_ALL_LIP2	0,0085	0,0084	0,0433	0,0445	0,0431	

BAYSNP	COMPARISON	GTYPE CPVAL	GTYPE XPVAL	ALLELE CPVAL	ALLELE XPVAL	ALLELE LRPVAL
1837	HELD_ALL_ADDRSULN	0,0159	0,015	0,0135	0,0245	0,0271
1837	HELD_MAL_ADR	0,0544	0,0558	0,0529	0,078	0,0897
1837	HELD_MAL_LIP2	0,0694	0,0696	0,0684	0,0215	0,0237
1870	HELD_ALL_CC	0,0213	0,018	0,0195	0,0874	0,1157
1870	HELD_FEM_CC	0,0621	0,0435	0,059	0,0937	0,1293
1882	CVD_MAL	0,0296	0,028	0,0055	0,4108	0,4529
1988	HELD_ALL_LIP	0,1287	0,1307	0,1234	0,0385	0,0414
2000	CVD_MAL	0,0237	0,0363	0,0295	0,0014	0,0025
2000	CVD_ALL	0,034	0,0425	0,035	0,0027	0,0035
2000	HELD_FBM_CC2	0,0705	0,0992	0,061	0,0105	0,0145
2000	HELD_MAL_HDL	0,1671	0,489	0,1018	0,0507	0,1177
2000	HELD_FEM_ADR	0,1624	0,2773	0,1528	0,0482	0,0704
2000	HELD_MAL_CC	0,1597	0,2882	0,1581	0,0467	0,063
2071	CVD_ALL	0,0823	0,09	0,0741	0,0349	0,0411
2078	HELD_MAL_LIP	0,0667	0,0395	0,0572	0,0468	0,0583
2085	HELD_FEM_VEFF	0,0707	0,0839	0,0347	0,019	0,0349
2095	CVD_ALL	0,0917	0,1451	0,0384	0,0935	0,1473
2119	HELD_MAL_LIP	0,0309	0,0409	0,0248	0,1269	0,148
2119	HELD_ALL_LIP	0,0382	0,0476	0,0373	0,133	0,1514
2119	HELD_FEM_EFF	0,057	0,0796	0,0527	0,1279	0,1563
2141	HELD_FEM_EFF	0,021	0,0256	0,0169	0,2401	0,3207

BAYSNP	COMPARISON	GTYPE CPVAL	GTYPE XPVAL	GTYPE LRPVAL	ALLELE CPVAL	ALLELE XPVAL	ALLELE LRPVAL
2141	HELD_ALL_CC	0,079	0,0695	0,0439	0,9551	1	0,9551
2182	HELD_FEM_EFF	0,0038	0,0027	0,0014	0,0039	0,0051	0,0033
2234	HELD_MAL_LIP	0,0604	0,0581	0,0195	0,0315	0,0414	0,0289
2281	HELD_FEM_VEFF	0,1098	0,1234	0,0542	0,0501	0,0685	0,0472
2298	CVD_FEM	0,0241	0,0171	0,0108	0,9341	1	0,934
2298	HELD_MAL_CC2	0,1235	0,1076	0,0833	0,053	0,0671	0,0514
2341	HELD_FEM_CC	0,0284	0,0709	0,0083	0,0336	0,0796	0,0097
2357	HELD_ALL_CC2	0,042	0,0374	0,016	0,7724	0,8793	0,7723
2357	HELD_ALL_CC	0,0452	0,0325	0,0209	0,9622	1	0,9622
2357	HELD_MAL_LIP	0,0438	0,0824	0,0385	0,077	0,1278	0,0657
2357	HELD_FEM_CC	0,0772	0,0829	0,0381	0,6486	0,7985	0,6469
2366	CVD_FEM	0,1125	0,1171	0,1073	0,0234	0,0304	0,023
2423	CVD_FEM	0,086	0,0888	0,077	0,0185	0,0274	0,0179
2708	CVD_FEM	0,0719	0,1262	0,054	0,0813	0,1384	0,0609
2995	HELD_FEM_ADDSULN	0,0882	0,0827	0,1088	0,0448	0,0488	0,0503
2995	HELD_FEM_UEFF	0,0943	0,0942	0,0928	0,0516	0,0693	0,0495
3360	HELD_MAL_ADDSULN	0,1131	0,1691	0,0302	0,0499	0,0819	0,0097
3464	HELD_ALL_CC	0,0305	0,0331	0,0278	0,0047	0,0056	0,0046
3464	HELD_FEM_CC	0,0743	0,0777	0,0721	0,0141	0,0184	0,0144
3689	HELD_FEM_EFF	0,0488	0,0584	0,0295	0,0226	0,0378	0,0206
3975	HELD_FEM_UEFF	0,0492	0,0474	0,0407	0,0198	0,0237	0,0188

BAYSNP	COMPARISON	GTYPE CPVAL	GTYPE XPVAL	GTYPE LRPVAL	ALLELE CPVAL	ALLELE XPVAL	ALLELE LRPVAL
3976	HELD_FEM_UEFF	0,059	0,0605	0,0456	0,0262	0,0327	0,025
4206	HELD_FEM_ADR3ULN	0,1395	0,1496	0,1372	0,0522	0,0655	0,0529
4838	HELD_FEM_VEFF	0,0581	0,0772	0,0529	0,0343	0,0681	0,0306
4838	HELD_FEM_VEFF	0,0581	0,0772	0,0529	0,0343	0,0681	0,0306
4838	HELD_FEM_VEFF	0,0581	0,0772	0,0529	0,0343	0,0681	0,0306
4912	HELD_FEM_EFF	0,1257	0,1748	0,0921	0,0255	0,0361	0,0255
4925	HELD_FEM_CC	0,0436	0,0623	0,0423	0,0685	0,0895	0,0679
4966	HELD_MAL_ADR3ULN	0,0269	0,0282	0,0298	0,1675	0,1966	0,1669
5014	HELD_ALL_ADR5ULN	0,007	0,0104	0,0022	0,0738	0,0869	0,0611
5014	HELD_FEM_ADR5ULN	0,0574	0,0604	0,0276	0,2347	0,2691	0,2164
5296	CVD_FEM	0,0459	0,0738	0,0438	0,0585	0,0899	0,0558
5296	HELD_FEM_EFF	0,0703	0,0489	0,0461	0,4109	0,5177	0,4006
5296	CVD_ALL	0,145	0,1027	0,1148	0,0579	0,0771	0,0523
5298	HELD_FEM_EFF	0,0813	0,0465	0,0567	0,4984	0,7366	0,49
5298	CVD_ALL	0,107	0,1065	0,0603	0,0348	0,0376	0,0306
5298	CVD_FEM	0,1629	0,1593	0,1332	0,0511	0,0885	0,049
5320	HELD_FEM_EFF	0,037	0,0397	0,029	0,016	0,0243	0,0151
5361	CVD_MAL	0,0947	0,1065	0,0447	0,0519	0,0654	0,0518
5457	HELD_FEM_EFF	0,1213	0,134	0,0452	0,2429	0,3056	0,2246
5704	HELD_MAL_LIP	0,0385	0,0334	0,0406	0,054	0,0678	0,0503
5704	CVD_MAL	0,0701	0,0755	0,07	0,0246	0,0281	0,0259

BAYSNP	COMPARISON	GTYPE CPVAL	GTYPE XPVAL	GTYPE LRPVAL	ALLELE CPVAL	ALLELE XPVAL	ALLELE LRPVAL
5717	HELD_FEM_ADR3ULN	0,0736	0,0775	0,0739	0,0219	0,026	0,021
5717	HELD_ALL_ADR3ULN	0,1246	0,1264	0,1214	0,0391	0,0471	0,0389
5959	HELD_ALL_CC	0,0126	0,0122	0,0098	0,0046	0,0073	0,0044
5959	CVD_FEM	0,019	0,0225	0,0082	0,0089	0,0137	0,0083
5959	HELD_MAL_CC	0,0525	0,0589	0,0243	0,0536	0,0708	0,053
5959	HELD_MAL_ADRSULN	0,038	0,0364	0,0482	0,1839	0,2158	0,1795
5959	HELD_FEM_ADR	0,054	0,0574	0,0527	0,0465	0,0539	0,0461
6162	HELD_ALL_ADR3ULN	0,0037	0,0034	0,0015	0,8524	0,9082	0,8522
6162	HELD_ALL_ADR	0,0033	0,003	0,0028	0,663	0,722	0,663
6162	HELD_ALL_ADRSULN	0,0206	0,0248	0,006	0,9797	1	0,9797
6162	HELD_MAL_ADR3ULN	0,0412	0,0352	0,0108	0,4721	0,4836	0,468
6162	HELD_FEM_ADRSULN	0,0274	0,0257	0,0147	0,4282	0,5487	0,4335
6162	HELD_MAL_ADR	0,0219	0,0217	0,0188	0,5399	0,6036	0,5399
6236	HELD_ALL_ADRSULN	0,0477	0,0396	0,0641	0,0131	0,016	0,0158
6236	HELD_MAL_ADR3ULN	0,0787	0,0734	0,0762	0,0279	0,0376	0,0305
6236	HELD_MAL_ADRSULN	0,0932	0,0861	0,0924	0,0297	0,0375	0,0368
6236	HELD_ALL_ADR3ULN	0,1516	0,1516	0,1604	0,0474	0,051	0,0497
6482	HELD_MAL_HDL	0,0359	0,0402	0,0326	0,009	0,013	0,0087
6482	HELD_ALL_LIP2	0,0383	0,0381	0,0383	0,0486	0,0506	0,0487
6482	HELD_MAL_CC2	0,0613	0,0667	0,0572	0,0114	0,0142	0,0106
6482	HELD_MAL_LIP2	0,0651	0,0662	0,065	0,0357	0,04	0,0358

BAYSNP	COMPARISON	GTYPE CPVAL	GTYPE XPVAL	ALLELE CPVAL	ALLELE XPVAL	ALLELE LRPVAL
6498	CVD_FEM	0,145	0,1987	0,0811	0,0323	0,0389
6744	HELD_ALL_ADDRESS	0,0659	0,07	0,0775	0,02	0,0273
7133	HELD_MAL_CC	0,0153	0,0278	0,0067	0,0006	0,0007
8021	CVD_FEM	0,039	0,0422	0,0304	0,8726	1
8060	CVD_FEM	0,044	0,0304	0,0304	0,1299	0,1961
8060	HELD_FEM_HDL	0,0558	0,0753	0,0549	0,0759	0,0965
8210	HELD_FEM_EFF	0,0336	0,0396	0,0276	0,3226	0,4454
8592	HELD_FEM_VEFF	0,0395	0,0432	0,0388	0,8842	0,9331
8816	HELD_FEM_EFF	0,0448	0,0448	0,0202	0,0144	0,0199
8846	HELD_ALL_LIP	0,0628	0,0654	0,0521	0,3798	0,3932
8943	HELD_MAL_LIP	0,1444	0,1408	0,0649	0,0389	0,0633
9193	HELD_FEM_LIP	0,0561	0,0723	0,0548	0,0707	0,0889
9193	CVD_FEM	0,1616	0,1289	0,1306	0,0458	0,0687
9443	CVD_MAL	0,0828	0,0869	0,0213	0,0507	0,0634
9516	HELD_MAL_CC	0,0504	0,0583	0,046	0,029	0,043
9698	HELD_MAL_ADDR	0,0106	0,0048	0,0061	0,0001	0,0001
9698	HELD_MAL_ADDRESS	0,0279	0,0274	0,0035	0,0003	0,0002
9698	HELD_FEM_EFF	0,0538	0,0557	0,0464	0,2251	0,2386
9698	HELD_MAL_ADDRESS	0,2515	0,3809	0,097	0,0239	0,0263
9698	CVD_ALL	0,2256	0,2237	0,2119	0,0274	0,0357

BAYSNP	COMPARISON	GTYPE CPVAL	GTYPE XPVAL	GTYPE LRPVAL	ALLELE CPVAL	ALLELE XPVAL	ALLELE LRPVAL
9849	HELD_FEM_CC	0,0302	0,0602	0,0168	0,0327	0,063	0,0182
9849	HELD_MAL_LIP	0,0315	0,0448	0,0358	0,0376	0,0505	0,043
9883	HELD_FEM_CC	0,006	0,0053	0,0046	0,6913	0,8398	0,6915
9883	HELD_ALL_CC	0,0345	0,035	0,0331	0,5629	0,6344	0,563
10079	CVD_ALL	0,118	0,0767	0,048	0,0611	0,0864	0,0418
10079	CVD_MAL	0,1491	0,2983	0,0682	0,0413	0,054	0,0099
10481	HELD_FEM_ADDR5ULN	0,0697	0,0667	0,0774	0,0136	0,0149	0,0135
10542	HELD_FEM_UEFF	0,0374	0,0214	0,0265	0,0981	0,1126	0,0911
10542	HELD_MAL_ADDR5ULN	0,1163	0,1946	0,0404	0,1357	0,2186	0,046
10600	HELD_FEM_EFF	0,0973	0,1483	0,0418	0,104	0,1554	0,0445
10621	HELD_FEM_CC	0,0622	0,0649	0,0451	0,373	0,4126	0,3769
10745	HELD_ALL_ADDR5ULN	0,0329	0,0356	0,0723	0,0754	0,0953	0,0832
10745	HELD_FEM_VEFF	0,0308	0,0308	0,0302	0,3022	0,3181	0,302
10747	HELD_MAL_ADR	0,006	0,0053	0,0044	0,6116	0,64	0,6115
10747	CVD_ALL	0,0285	0,0292	0,027	0,1252	0,1349	0,1253
10747	HELD_MAL_ADDR3ULN	0,0401	0,0412	0,0505	0,8735	1	0,8734
10771	HELD_MAL_ADDR5ULN	0,0176	0,0191	0,0469	0,0263	0,0458	0,0291
10771	HELD_FEM_EFF	0,1837	0,1844	0,1832	0,0527	0,0543	0,0525
10870	HELD_MAL_LIP	0,0323	0,0272	0,0156	0,8328	1	0,8332
10870	HELD_FEM_LIP	0,0431	0,0412	0,0421	0,0319	0,037	0,0317
10870	HELD_MAL_CC	0,1157	0,0954	0,0779	0,0341	0,0413	0,0285

BAYSNP	COMPARISON	GTYPE CPVAL	GTYPE XPVAL	GTYPE LRPVAL	ALLELE CPVAL	ALLELE XPVAL	ALLELE LRPVAL
10870	HELD_ALL_CC	0,1146	0,1205	0,109	0,0272	0,0351	0,027
10877	HELD_ALL_HDL	0,0907	0,1181	0,0333	0,0266	0,0356	0,007
10948	HELD_FEM_LIP	0,0134	0,0136	0,0127	0,052	0,0588	0,0517
10948	HELD_ALL_LIP	0,0209	0,0207	0,0197	0,0356	0,0432	0,0355
10948	HELD_FEM_CC2	0,0513	0,0521	0,0493	0,3385	0,3602	0,3382
10948	CVD_MAL	0,0986	0,0986	0,103	0,0481	0,0548	0,0475
11001	HELD_MAL_ADDSULN	0,0438	0,0618	0,1215	0,1034	0,1201	0,1152
11073	HELD_MAL_ADDSULN	0,1741	0,1866	0,1892	0,0446	0,0632	0,0503
11153	HELD_FEM_CC	0,0378	0,0459	0,038	0,064	0,0726	0,0658
11210	HELD_MAL_CC	0,025	0,0616	0,0225	0,0335	0,0756	0,0304
11210	HELD_ALL_ADD3ULN	0,0344	0,027	0,0311	0,076	0,0917	0,0844
11210	HELD_ALL_ADR	0,0536	0,038	0,0354	0,2211	0,2468	0,2195
11248	HELD_FEM_ADR	0,0125	0,0119	0,0118	0,0368	0,0494	0,0364
11248	HELD_MAL_LIP	0,0478	0,0677	0,0404	0,0784	0,1038	0,0644
11248	HELD_ALL_CC	0,0431	0,0567	0,0425	0,0874	0,1066	0,0887
11372	HELD_MAL_LIP	0,2326	0,2665	0,2343	0,0486	0,0753	0,0477
11449	HELD_FEM_CC	0,0245	0,0119	0,0204	0,0644	0,0971	0,0663
11450	HELD_FEM_EFF	0,0922	0,0949	0,0903	0,0362	0,0394	0,036
11470	HELD_MAL_LIP	0,0807	0,1484	0,0304	0,0882	0,1582	0,033
11472	HELD_MAL_LIP	0,0763	0,1465	0,0284	0,0836	0,1565	0,031
11472	HELD_FEM_LIP	0,0576	0,0991	0,0495	0,0617	0,1046	0,053

BAYSNP	COMPARISON	GTYPE CPVAL	GTYPE XPVAL	GTYPE LRPVAL	GTYPE CPVAL	ALLELE XPVAL	ALLELE LRPVAL
11487	HELD_MAL_ADR5ULN	0,0033	0,0039	0,0004	0,0122	0,0159	0,0012
11487	HELD_MAL_ADR3ULN	0,0156	0,021	0,0131	0,038	0,0474	0,0295
11488	HELD_MAL_ADR5ULN	0,0117	0,0227	0,0018	0,0076	0,0087	0,0006
11488	HELD_FEM_UEFF	0,0217	0,021	0,0091	0,0655	0,0713	0,0672
11488	HELD_MAL_ADR3ULN	0,0239	0,0311	0,0166	0,0898	0,127	0,0797
11493	HELD_MAL_CC	0,0736	0,0542	0,0493	0,6283	0,7502	0,6293
11502	HELD_MAL_ADR3ULN	0,0881	0,0865	0,0363	0,0283	0,0301	0,0225
11502	HELD_MAL_ADR5ULN	0,1706	0,154	0,1118	0,0592	0,0659	0,0396
11534	HELD_ALL_LIP	0,1034	0,2501	0,0513	0,1046	0,2518	0,0519
11537	CVD_FEM	0,1061	0,1119	0,0989	0,0221	0,0256	0,0214
11537	HELD_FEM_EFF	0,1916	0,2436	0,1166	0,0438	0,0655	0,0324
11560	HELD_FEM_EFF	0,1693	0,3529	0,1436	0,0519	0,1212	0,0386
11578	HELD_FEM_LIP	0,0201	0,0333	0,0132	0,0226	0,0366	0,0147
11578	CVD_FEM	0,0435	0,0775	0,0229	0,0459	0,0799	0,0241
11594	HELD_FEM_ADR3ULN	0,1373	0,2125	0,0418	0,0279	0,0331	0,0052
11594	HELD_ALL_ADR5ULN	0,1669	0,1552	0,0434	0,0516	0,0536	0,0092
11594	HELD_ALL_CC	0,0539	0,0724	0,0479	0,0648	0,0846	0,0574
11594	HELD_ALL_ADR	0,1052	0,0878	0,1	0,0304	0,036	0,0286
11594	HELD_FEM_ADR5ULN	0,3753	0,4458	0,1824	0,1236	0,213	0,0409
11624	HELD_ALL_CC	0,0352	0,0383	0,0111	0,3119	0,388	0,3111
11624	HELD_MAL_CC	0,032	0,0313	0,0164	0,6153	0,7739	0,6163

BAYSNP	COMPARISON	GTYPE CPVAL	GTYPE XPVAL	GTYPE LRPVAL	ALLELE CPVAL	ALLELE XPVAL	ALLELE LRPVAL
11624	HELD_FEM_EFF	0,2292	0,244	0,1389	0,053	0,0656	0,0407
11627	HELD_ALL_CC	0,0337	0,0316	0,0088	0,0936	0,1309	0,0921
11627	HELD_MAL_CC	0,0931	0,0933	0,0528	0,352	0,4146	0,3531
11627	HELD_FEM_EFF	0,1916	0,2436	0,1166	0,0438	0,0655	0,0324
11644	HELD_MAL_DR5ULN	0,2097	0,2525	0,1344	0,0676	0,1027	0,0467
11650	HELD_FEM_EFF	0,0366	0,0361	0,0363	0,1123	0,1212	0,1122
11654	HELD_ALL_DR5ULN	0,0052	0,0046	0,0042	0,6623	0,7404	0,6642
11654	HELD_FEM_DR5ULN	0,0104	0,0096	0,006	0,7072	0,832	0,7087
11654	HELD_FEM_ADDR3ULN	0,0546	0,0592	0,0524	0,6906	0,7512	0,6913
11654	HELD_ALL_ADDR3ULN	0,052	0,0518	0,0601	0,2706	0,2742	0,2735
11655	HELD_ALL_DR5ULN	0,0085	0,0074	0,0058	0,8555	0,8723	0,8558
11655	HELD_FEM_DR5ULN	0,0136	0,0138	0,0053	0,7681	0,8443	0,7672
11655	HELD_FEM_ADDR3ULN	0,0489	0,048	0,0432	0,9169	1	0,9169
11656	HELD_MAL_LIP	0,0321	0,0317	0,0346	0,012	0,0141	0,0126
11656	HELD_FEM_EFF	0,0782	0,0909	0,0511	0,0442	0,0652	0,0393
11656	HELD_ALL_LIP	0,0617	0,0646	0,06	0,0295	0,0353	0,0295
11825	HELD_MAL_DR5ULN	0,0233	0,056	0,0499	0,0278	0,0619	0,0612
11914	HELD_MAL_DR5ULN	0,0186	0,0915	0,0128	0,0001	0,0001	0
11914	HELD_ALL_DR5ULN	0,1572	0,1781	0,1391	0,0477	0,0533	0,0487
12008	HELD_FEM_EFF	0,0222	0,0317	0,0209	0,0249	0,0351	0,0234
12008	HELD_ALL_DR5ULN	0,1272	0,2155	0,0422	0,135	0,225	0,0445

BAYSNP	COMPARISON	GTYPE CPVAL	GTYPE LRPVAL	ALLELE CPVAL	ALLELE LRPVAL
12097	HELD_ALL_ADDR5ULN	0,0162	0,0277	0,0308	0,019
12097	HELD_FEM_ADDR3ULN	0,0342	0,0487	0,042	0,0392
12097	HELD_MAL_ADDR5ULN	0,04	0,0749	0,0726	0,0462
12097	HELD_ALL_ADDR3ULN	0,0465	0,073	0,056	0,0524
12366	HELD_FEM_UEFF	0,0342	0,0313	0,0069	0,0364
12366	HELD_ALL_ADDR5ULN	0,0464	0,0391	0,0411	0,5197
12619	HELD_MAL_ADDR5ULN	0,0073	0,1235	0,0387	0,0075
12619	HELD_ALL_ADDR5ULN	0,0121	0,0605	0,0414	0,0125
13025	HELD_ALL_ADDR5ULN	0,04	0,0399	0,0593	0,3978
13191	HELD_FEM_LIP	0,0157	0,0149	0,015	0,0072
13191	HELD_ALL_ADDR5ULN	0,0648	0,0601	0,0431	0,0199
13191	HELD_ALL_LIP	0,0634	0,0669	0,0616	0,0211
13937	HELD_FEM_ADDR5ULN	0,076	0,0835	0,0789	0,0402
900002	CVD_FEM	0,1492	0,1674	0,1456	0,0364
900013	CVD_FEM	0,0212	0,022	0,0192	0,2613
900013	CVD_ALL	0,0279	0,0289	0,0279	0,1847
900025	CVD_MAL	0,1379	0,1533	0,1361	0,0426
900032	CVD_FEM	0,0555	0,036	0,0317	0,2549
900045	HELD_FEM_EFF	0,162	0,2388	0,151	0,0411
900065	CVD_FEM	0,0222	0,0175	0,0086	0,0066
900065	CVD_MAL	0,0549	0,0421	0,0289	0,4512

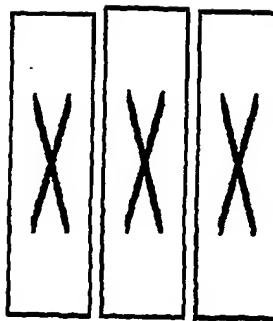
BAYSNP	COMPARISON	GTYPE CPVAL	GTYPE XPVAL	GTYPE LRPVAL	ALLELE CPVAL	ALLELE XPVAL	ALLELE LRPVAL
900065	CVD_ALL	0,0773	0,0753	0,0754	0,0471	0,0505	0,0477
900078	HELD_ALL_ADR3ULN	0,0283	0,036	0,0348	0,0335	0,0417	0,0415
900078	HELD_ALL_ADR5ULN	0,03	0,0417	0,0487	0,0349	0,0466	0,0574
900078	HELD_FEM_ADR3ULN	0,0342	0,0487	0,042	0,0392	0,0543	0,0484
900082	HELD_FEM_ADR3ULN	0,0377	0,0378	0,0364	0,01073	0,111	0,1055
900082	HELD_FEM_ADR5ULN	0,0517	0,0587	0,0566	0,0581	0,0837	0,0542
900096	CVD_ALL	0,0644	0,0622	0,0602	0,032	0,0354	0,0294
900107	HELD_MAL_ADR5ULN	0,2371	0,2767	0,1405	0,0665	0,1045	0,0455
900115	HELD_MAL_ADR5ULN	0,0214	0,02	0,0409	0,0148	0,0208	0,0158
900115	HELD_FEM_EFF	0,0347	0,0338	0,0316	0,4668	0,5083	0,4661
900121	HELD_MAL_ADR	0,0303	0,0297	0,0268	0,3005	0,3162	0,3003
900173	CVD_ALL	0,1397	0,146	0,1347	0,0356	0,0569	0,0349
1000002	HELD_FEM_EFF	0,0781	0,0766	0,0305	0,0098	0,0139	0,0067
1000006	HELD_FEM_CC	0,0041	0,0018	0,0035	0,0014	0,0024	0,0014
1000006	HELD_ALL_CC	0,0127	0,0087	0,0113	0,0023	0,0034	0,002
1000014	HELD_ALL_CC	0,0156	0,0099	0,013	0,0468	0,0612	0,046
1000014	HELD_FEM_CC	0,0415	0,0248	0,0336	0,1157	0,1943	0,1184
1000025	HELD_MAL_LIP	0,1055	0,1309	0,0337	0,1763	0,2188	0,1719

Table 6a Correlation of genotypes of PA SNPs to relative risk

For diagnostic conclusions to be drawn from genotyping a particular patient we calculated the relative risk RR1, RR2, RR3 for the three possible genotypes of each SNP. Given the genotype frequencies as

	gtype1	gtype2	gtype3
case	N11	N12	N13
control	N21	N22	N23

we calculate



Here, the *case* and *control* populations represent any case-control-group pair, or bad(case)-good(control)-group pair, respectively (due to their increased response to statins, 'high responders' are treated as a case cohort, whereas 'low responders' are treated as the respective control cohort). A value $RR1>1$, $RR2>1$, and $RR3>1$ indicates an increased risk for individuals carrying genotype 1, genotype 2, and genotype 3, respectively. For example, $RR1=3$ indicates a 3-fold risk of an individual carrying genotype 1 as compared to individuals carrying genotype 2 or 3 (a detailed description of relative risk calculation and statistics can be found in (Biostatistics, L. D. Fisher and G. van Belle, Wiley Interscience 1993)). The baySNP number refers to an internal numbering of the PA SNPs and can be found in the sequence listing. null: not defined.

In cases where a relative risk is not given in the table (three times zero or null) the informative genotype can be drawn from the right part of the table where the frequencies of genotypes are given in the cases and control cohorts. For example BaySNP 3360 gave the following results:

BAYSNP	COMPARISON	GTYPE1	GTYPE2	GTYPE3	RR1	RR2	RR3
	HELD_MAL_ADDRSULN	GG	GT	TT	null	0	0
3360							

FQ1_A	FQ2_A	FQ3_A	FQ1_B	FQ2_B	FQ3_B
10	0	0	50	22	1

It can be concluded that a GT or TT genotype is only present in the control cohort; these genotypes are somehow protective against ADR. An analogous proceeding can be used to determine protective alleles if no relative risk is given (table 6b).

BAYSNP	COMPARISON	GTYPE1	GTYPE2	GTYPE3	RRI	RR2	RR3	FQ1_A	FQ2_A	FQ3_A	FQ1_B	FQ2_B	FQ3_B
28	HELD_FEM_EFF	CC	CT	TT	0,68	0,29	3,38	1	2	9	3	12	7
29	HELD_ALL_HDL	AA	AG	GG	0	0,90	0,58	4	4	2	0	7	8
29	HELD_MALADR3ULN	AA	AG	GG	2,16	0,56	0,75	13	7	6	18	32	22
29	HELD_MALADR5ULN	AA	AG	GG	3,15	0,66	0,32	5	3	1	18	32	22
52	HELD_FEM_EFF	CC	CG	GG	1,96	1,02	0,23	7	10	1	5	17	9
56	HELD_FEM_BFF	AA	AG	GG	null	2,76	0,36	0	5	7	0	2	20
89	HELD_ALL_CC	AA	AG	null	null	0	null	45	0	0	37	3	0
90	HELD_FEM_CC	CC	CT	TT	0,97	0,64	1,82	8	13	10	6	15	1
99	HELD_FEM_LIP	CC	CT	TT	1,51	0,7	1,16	13	28	41	5	41	34
140	HELD_FEM_EFF	CC	CT	TT	0	0	null	0	0	12	1	2	18
152	HELD_FEM_EFF	AA	AG	GG	0,42	1,27	2,5	3	6	3	12	9	1
214	HELD_ALL_LIP	AA	AG	GG	0,92	1,18	0	59	38	0	73	36	4
214	HELD_FEM_LIP	AA	AG	GG	1	1,11	0	50	31	0	48	26	4
221	HELD_ALL_CC	CC	CG	GG	1,36	0,56	1,44	7	12	26	3	21	15
221	HELD_FEM_CC	CC	CG	GG	1,16	0,53	1,67	4	9	18	2	14	6
224	HELD_FEM_LIP	CC	CT	TT	0,77	1,26	1,24	51	8	20	60	5	14
224	HELD_MAL_LIP	CC	CT	TT	2,02	1,45	0,38	17	1	2	25	1	11
294	HELD_ALL_CC	CC	CT	TT	0,83	0,97	2	16	24	5	18	22	0
307	CVD_FEM	CC	CT	TT	0,34	0,8	1,84	2	15	19	9	20	9
307	HELD_ALL_LIP	CC	CT	TT	null	1,41	0,71	0	70	32	0	63	54
411	HELD_ALL_HDL	AA	AT	TT	1,85	0,69	0,56	7	3	0	5	8	2

BAYSNP	COMPARISON	GTYPE1	GTYPE2	GTYPE3	RR1	RR2	RR3	FQ1_A	FQ2_A	FQ3_A	FQ1_B	FQ2_B	FQ3_B
449	HELD_MAL_LIP	CC	CG	GG	0	0,42	2,62	0	3	17	1	14	22
466	CVD_FEM	CC	CT	TT	0,66	0,86	1,61	6	15	14	12	20	8
472	HELD_FEM_EFF	AA	AG	GG	0	0	null	0	0	11	3	6	13
542	HELD_MAL_CC	AA	AG	GG	2,58	3,07	0,23	2	8	4	0	2	17
542	HELD_MAL_HDL	AA	AG	GG	0	2,38	0,30	3	8	10	0	3	24
542	HELD_ALL_ADR	AA	AG	GG	0	1,32	0,78	0	53	106	2	33	119
542	HELD_FEM_HDL	AA	AG	GG	0,57	0,67	1,56	0	2	21	1	8	23
739	HELD_ALL_CC	CC	CG	GG	0,67	0,94	1,52	9	21	15	14	20	6
821	HELD_MAL_LIP2	AA	AC	CC	1,4	0,96	0,93	32	116	161	18	138	193
821	HELD_FEM_VEFF	AA	AC	CC	0	0,93	2,1	0	4	6	4	6	4
1005	HELD_MAL_CC	AA	AG	GG	2,35	0,6	0	12	2	0	11	5	2
1055	HELD_MAL_CC	AA	AT	TT	0	3	1	0	3	6	4	0	8
1056	HELD_FEM_EFF	AA	AG	GG	1,59	0,37	2,04	12	6	6	10	21	2
1085	HELD_MAL_LIP	AA	AG	GG	0,37	1,31	1,75	3	11	6	15	16	5
1085	CVD_FEM	AA	AG	GG	1,51	0,88	0,5	20	11	3	16	15	9
1086	HELD_MAL_LIP	AA	AG	GG	1,97	1	0,44	7	10	3	5	18	13
1092	HELD_MAL_LIP	CC	CG	GG	0,94	0,4	2,38	2	5	13	4	21	12
1096	HELD_MAL_CC	GG	GT	TT	null	2,2	0,45	0	7	7	0	3	15
1096	CVD_MAL	GG	GT	TT	1,51	0,72	1,22	4	13	52	0	12	21
1101	HELD_FEM_EFF	CC	CT	TT	null	0	null	12	0	0	18	4	0
1204	HELD_MAL_LIP	AA	AG	GG	3,06	1,58	0,49	2	8	9	0	9	26
1204	HELD_ALL_LIP	AA	AG	GG	1,34	1,18	0,77	12	38	49	8	36	71

BAYSNP	COMPARISON	GTYPE1	GTYPE2	GTYPE3	RRI	RR2	RR3	FQ1_A	FQ2_A	FQ3_A	FQ1_B	FQ2_B	FQ3_B
1504	HELD_ALL_CC	CC	CT	TT	0,5	1,79	0,78	5	27	12	12	12	15
1504	HELD_MAL_LIP	CC	CT	TT	0	1,6	1,14	0	12	7	8	17	12
1504	HELD_MAL_CC	CC	CT	TT	0,72	2,63	0,4	2	9	3	4	4	10
1504	HELD_FEM_CC	CC	CT	TT	0,4	1,44	1,13	3	18	9	8	8	5
1511	HELD_FEM_EFF	GG	GT	TT	0,33	3,38	0	3	9	0	14	7	1
1524	HELD_FEM_ADDR3ULN	AA	AC	CC	0	1,51	0,89	0	16	22	8	23	51
1556	HELD_FEM_EFF	CC	CG	GG	null	3,36	0,3	0	7	5	0	3	19
1561	CVD_FEM	AA	AC	CC	1,59	0,73	0,41	23	12	1	17	19	4
1582	HELD_MAL_LIP	CC	CT	TT	0	0,78	1,89	0	5	15	5	12	20
1638	HELD_FEM_CC	AA	AG	GG	0,56	0,62	1,73	1	8	22	2	11	9
1653	CVD_MAL	GG	GT	TT	0,86	1,43	0,71	15	40	14	10	10	13
1662	HELD_MAL_CC	CC	CT	TT	2,8	null	0,36	4	0	10	0	0	18
1714	CVD_MAL	AA	AG	GG	0,48	0,98	1,23	3	26	37	6	14	14
1722	HELD_FEM_ADDR5ULN	CC	CT	TT	2,8	0,41	0,93	8	5	5	14	43	24
1757	HELD_FEM_EFF	AA	AG	GG	3	0,68	0,88	4	7	9	0	16	16
1765	HELD_ALL_ADDR3ULN	AA	AG	GG	0,67	0,36	2,71	1	7	55	4	48	97
1765	HELD_ALL_ADDR3ULN	AA	AG	GG	0,67	0,36	2,71	1	7	55	4	48	97
1765	HELD_ALL_ADDRSULN	AA	AG	GG	null	0,31	3,64	0	3	24	4	48	97
1765	HELD_MAL_ADDR3ULN	AA	AG	GG	0	0,26	4,23	0	2	24	2	21	47
1765	HELD_MAL_ADDR3ULN	AA	AG	GG	0	0,26	4,23	0	2	24	2	21	47
1765	HELD_MAL_ADDRSULN	AA	AG	GG	0	0	null	0	0	10	-	2	21

BAYSNP	COMPARISON	GTYPE1	GTYPE2	GTYPE3	RRI	RR2	RR3	FQ1_A	FQ2_A	FQ3_A	FQ1_B	FQ2_B	FQ3_B
1765	HELD_MAL_ADRSULN	AA	AG	GG	0	0	null	0	0	10	2	21	47
1765	HELD_FEM_ADR3ULN	AA	AG	GG	1,05	0,41	2,23	1	5	31	2	27	50
1765	HELD_FEM_ADR3ULN	AA	AG	GG	1,05	0,41	2,23	1	5	31	2	27	50
1776	HELD_ALL_CC	AA	AG	GG	null	null	0	45	0	0	37	0	3
1776	HELD_FEM_CC	AA	AG	GG	null	null	0	31	0	0	20	0	2
1799	HELD_FEM_LIP2	CC	CT	TT	1,04	0,82	1,4	123	119	49	145	178	33
1799	HELD_MAL_CC	CC	CT	TT	0,45	1,46	1,91	4	7	3	11	6	1
1806	HELD_FEM_BFF	AA	AG	GG	3,96	0,35	0	11	1	0	14	6	2
1837	HELD_FEM_LIP2	CC	CT	TT	1,17	0,77	1,32	164	108	32	166	167	22
1837	HELD_ALL_LIP2	CC	CT	TT	1,18	0,83	1,04	334	223	50	322	308	52
1837	HELD_ALL_ADR5ULN	CC	CT	TT	2,82	0,34	0,86	20	6	2	66	76	13
1837	HELD_MAL_ADR	CC	CT	TT	1,45	0,7	0,96	37	33	7	21	44	7
1837	HELD_MAL_LIP2	CC	CT	TT	1,19	0,89	0,77	170	115	18	156	141	30
1870	HELD_ALL_CC	CC	CT	TT	0,73	1,75	0,61	2	25	18	3	10	26
1870	HELD_FEM_CC	CC	CT	TT	0,85	1,75	0,58	1	20	10	1	7	14
1882	CVD_MAL	CC	CT	TT	1,06	0,76	1,59	21	37	11	9	25	0
1988	HELD_ALL_LIP	CC	CT	TT	1,26	0,95	0,64	52	39	9	48	48	20
2000	CVD_MAL	CC	TT	null	2,45	0,41	null	68	2	0	29	5	0
2000	CVD_ALL	CC	TT	null	1,98	0,51	null	101	4	0	65	9	0
2000	HELD_FEM_CC2	CC	TT	null	3,29	0,3	null	45	1	0	37	5	0
2000	HELD_MAL_HDL	CC	TT	null	2,00	0,50	0	20	0	0	20	2	0
2000	HELD_FEM_ADR	CC	TT	null	2,01	0,5	null	77	2	0	76	6	0

BAYSNP	COMPARISON	GTYPE1	GTYPE2	GTYPE3	RRI	RR2	RR3	FQ1_A	FQ2_A	FQ3_A	FQ1_B	FQ2_B	FQ3_B
2000	HELD_MAL_CC	CC	TT	null	0,51	1,98	null	11	3	0	18	1	0
2071	CVD_ALL	AA	AG	GG	1,4	1,09	0,79	14	52	36	4	34	36
2078	HELD_MAL_LIP	GG	GT	TT	3,06	1,9	0,45	1	11	6	0	13	22
2085	HELD_FEM_VEFF	GG	GT	TT	2,5	0,79	0	6	4	0	3	7	4
2095	CVD_ALL	AG	GG	null	1,72	0,58	null	4	101	0	0	73	0
2119	HELD_MAL_LIP	AA	AG	null	0,35	2,83	null	3	17	0	16	21	0
2119	HELD_ALL_LIP	AA	AG	null	0,72	1,39	null	29	73	0	49	68	0
2119	HELD_FEM_EFF	AA	AG	null	0,38	2,67	null	3	9	0	13	9	0
2141	HELD_FEM_EFF	AA	AG	GG	0	3,25	0,42	0	6	6	2	2	18
2141	HELD_ALL_CC	AA	AG	GG	0	1,35	0,87	0	17	28	3	9	27
2182	HELD_FEM_EFF	AA	AG	GG	3,71	0,65	0	6	6	0	1	14	6
2234	HELD_MAL_LIP	GG	GT	TT	0	0,96	1,75	0	10	10	7	18	10
2281	HELD_FEM_VEFF	AA	AC	CC	0	1,04	2,13	0	5	4	4	7	2
2298	CVD_FEM	AA	AC	CC	2,23	0,57	1,31	4	10	21	0	20	18
2298	HELD_MAL_CC2	AA	AC	CC	0	0,7	1,65	0	8	21	2	12	14
2341	HELD_FEM_CC	CC	CT	TT	null	1,88	0,53	0	6	25	0	0	22
2357	HELD_ALL_CC2	AA	AG	GG	2,03	0,76	1,1	5	18	51	0	25	46
2357	HELD_ALL_CC	AA	AG	GG	1,98	0,62	1,21	4	8	33	0	14	26
2357	HELD_MAL_LIP	AA	AG	GG	0,42	2,4	0	4	4	16	0	17	19
2357	HELD_FEM_CC	AA	AG	GG	1,81	0,57	1,13	4	4	23	0	7	15
2366	CVD_FEM	GG	GT	TT	1,51	1,12	0,55	12	14	7	8	15	17
2423	CVD_FEM	AA	AG	GG	1,48	1,08	0,45	16	13	4	12	14	13

BAYSNP	COMPARISON	GTYPE1	GTYPE2	GTYPE3	RR1	RR2	RR3	FQ1_A	FQ2_A	FQ3_A	FQ1_B	FQ2_B	FQ3_B
2708	CVD_FEM	CC	CT	TT	3,67	0,27	null	28	1	0	33	7	0
2995	HELD_FEM_ADRSULN	AA	AC	CC	2,66	1,41	0,45	3	10	5	4	37	41
2995	HELD_FEM_VEFF	AA	AC	CC	0,67	0,68	1,57	2	20	32	5	40	30
3360	HELD_MAL_ADRSULN	GG	GT	TT	null	0	0	10	0	0	50	22	1
3464	HELD_ALL_CC	AA	AG	GG	0,43	0,83	1,61	3	15	27	9	17	14
3464	HELD_FEM_CC	AA	AG	GG	0,6	0,67	1,74	3	7	21	5	9	8
3689	HELD_FEM_EFF	CC	CG	GG	4	0,82	0	3	3	0	1	8	5
3975	HELD_FEM_VEFF	AA	AC	CC	0,37	0,83	1,5	2	24	30	10	38	27
3976	HELD_FEM_VEFF	AA	AG	GG	0,34	0,92	1,41	2	24	30	11	35	29
4206	HELD_FEM_ADR3ULN	AA	AT	TT	0,57	1,14	1,61	8	20	9	31	41	11
4838	HELD_FEM_VEFF	AA	AG	GG	3,27	0,35	0,56	7	2	1	3	8	3
4838	HELD_FEM_VEFF	AA	AG	GG	3,27	0,35	0,56	7	2	1	3	8	3
4838	HELD_FEM_VEFF	AA	AG	GG	3,27	0,35	0,56	7	2	1	3	8	3
4912	HELD_FEM_EFF	AA	AG	GG	2,33	0	0,56	7	0	5	5	2	13
4925	HELD_MAL_CC	AA	AC	CC	0,45	2,2	null	7	7	0	15	3	0
4966	HELD_MAL_ADR3ULN	AA	AG	GG	1,08	0,44	2,26	7	8	11	18	41	13
5014	HELD_ALL_ADRSULN	AA	AG	GG	1,54	0,16	3,07	3	2	23	10	57	85
5014	HELD_FEM_ADRSULN	AA	AG	GG	1,64	0,15	2,73	2	1	15	5	27	49
5296	CVD_FEM	AA	AG	GG	null	1,7	0,59	0	10	26	0	4	36
5296	HELD_FEM_EFF	AA	AG	GG	3	0,22	2,39	1	1	10	0	9	13
5296	CVD_ALL	AA	AG	GG	1,72	1,29	0,76	1	25	78	0	10	64
5298	HELD_FEM_EFF	CC	CT	TT	3,2	0,23	2,25	1	1	9	0	9	13

BAYSNP	COMPARISON	GTYPE1	GTYPE2	GTYPE3	RR1	RR2	RR3	FQ1_A	FQ2_A	FQ3_A	FQ1_B	FQ2_B	FQ3_B
5298	CVD_ALL	CC	CT	TT	1,76	1,24	0,76	3	22	76	0	10	64
5298	CVD_FEM	CC	CT	TT	2,18	1,56	0,61	1	8	26	0	4	36
5320	HELD_FEM_EFF	AA	AG	GG	0,23	0,88	2,18	1	10	8	9	19	5
5361	CVD_MAL	AA	AC	CC	0,77	1,54	1,16	24	5	35	18	0	14
5457	HELD_FEM_EFF	AA	AG	GG	1,41	0	3,52	1	0	11	1	6	14
5704	HELD_MAL_LIP	CC	CT	TT	0,7	0,45	2,44	1	8	11	3	26	8
5704	CVD_MAL	CC	CT	TT	0,65	0,87	1,32	5	30	33	6	18	9
5717	HELD_FEM_ADDR3ULN	AA	AG	GG	1,77	0,82	0,55	17	16	5	21	41	21
5717	HELD_ALL_ADDR3ULN	AA	AG	GG	1,44	1,01	0,64	21	32	12	34	76	46
5959	HELD_ALL_CC	AA	AG	GG	1,81	0,85	0,59	16	20	7	4	21	13
5959	CVD_FEM	AA	AG	GG	3,6	0,8	0,27	4	4	1	0	7	6
5959	HELD_MAL_CC	AA	AG	GG	2,7	0,82	0,57	4	7	3	0	10	7
5959	HELD_ALL_ADDR5ULN	AA	AG	GG	1,16	0,22	4,03	2	2	5	13	41	13
5959	HELD_FEM_ADR	AA	AG	GG	1,15	1,32	0,62	15	41	16	11	29	28
6162	HELD_ALL_ADDR3ULN	CC	CG	GG	0,15	1,78	0,77	1	35	28	19	52	80
6162	HELD_ALL_ADR	CC	CG	GG	0,45	1,33	0,9	6	76	74	19	52	80
6162	HELD_ALL_ADDR5ULN	CC	CG	GG	0	2,35	0,66	0	16	11	19	52	80
6162	HELD_MAL_ADDR3ULN	CC	CG	GG	0	1,85	0,87	0	13	13	11	21	39
6162	HELD_FEM_ADDR5ULN	CC	CG	GG	0	3,19	0,43	0	13	5	8	31	41
6162	HELD_MAL_ADR	CC	CG	GG	0,4	1,39	0,91	3	34	37	11	21	39
6236	HELD_ALL_ADDR5ULN	CC	CT	TT	2,41	1,25	0,49	6	12	9	13	58	81
6236	HELD_MAL_ADDR3ULN	CC	CT	TT	1,74	1,63	0,47	4	15	8	5	28	39

BAYSNP	COMPARISON	GTYPE1	GTYPE2	GTYPE3	RRI	RR2	RR3	FQ1_A	FQ2_A	FQ3_A	FQ1_B	FQ2_B	FQ3_B
6236	HELD_MAL_ADD5ULN	CC	CT	TT	2,68	2,12	0,25	2	6	2	5	28	39
6236	HELD_ALL_ADD3ULN	CC	CT	TT	1,58	1,15	0,71	10	27	26	13	58	81
6482	HELD_MAL_HDL	AA	AG	GG	0,44	1,96	1,79	5	8	4	15	4	2
6482	HELD_ALL_LIP2	AA	AG	GG	0,87	1,16	1	340	238	41	436	226	47
6482	HELD_MAL_CC2	AA	AG	GG	1,93	0,66	0,47	18	7	2	10	12	6
6482	HELD_MAL_LIP2	AA	AG	GG	0,83	1,2	1,08	173	115	21	220	99	20
6498	CVD_FEM	AA	AG	GG	1,85	0,73	0	28	4	0	25	7	3
6744	HELD_ALL_ADD5ULN	CC	CT	TT	2,27	1,54	0,47	4	13	9	9	56	84
7133	HELD_MAL_CC	CC	CG	GG	0,36	null	2,8	10	0	4	18	0	0
8021	CVD_FEM	AA	AG	GG	0,71	1,98	0,26	8	19	1	15	14	7
8060	CVD_FEM	AA	AG	GG	2,1	0,38	2,18	31	3	1	28	12	0
8060	HELD_FEM_HDL	AA	AG	GG	0,47	2,13	0	11	7	0	20	3	0
8210	HELD_FEM_EFF	AA	AG	GG	0,22	2,93	0,81	1	7	4	9	4	9
8592	HELD_FEM_VEFF	CC	CT	TT	0,7	1,32	0,86	15	92	43	25	68	50
8816	HELD_FEM_EFF	CC	CG	GG	2,22	1,17	0,36	4	7	2	0	5	6
8846	HELD_ALL_LIP	AA	AG	GG	1	1,18	0,4	57	47	3	62	42	12
8943	HELD_MAL_LIP	AA	AC	CC	1,89	0,78	0	15	5	0	20	12	5
9193	HELD_FEM_LIP	CC	CG	GG	1,54	0,65	null	72	11	0	60	20	0
9193	CVD_FEM	CC	CG	GG	0,59	1,59	2,14	28	7	1	37	3	0
9443	CVD_MAL	CC	CT	TT	1,55	1	0,85	9	25	35	0	12	21
9516	HELD_MAL_CC	AA	AG	GG	2,56	0,52	0,67	7	3	4	2	8	8
9698	HELD_MAL_ADDR	AA	AG	GG	0,41	0	2,78	4	0	70	14	2	56

BAYSNP	COMPARISON	GTYPE1	GTYPE2	GTYPE3	RRI	RR2	RR3	FQL_A	FQ2_A	FQ3_A	FQ1_B	FQ2_B	FQ3_B
9698	HELD_MAL_ADDR3ULN	AA	AG	GG	0	0	0	0	27	14	2	56	
9698	HELD_FEM_BFF	AA	AG	GG	0,47	1,04	1,04	5	95	194	16	91	191
9698	HELD_MAL_ADDR5ULN	AA	AG	GG	0	0	null	0	0	10	14	2	56
9698	CVD_ALL	AA	AG	GG	1,31	1,09	0,8	17	12	73	6	7	59
9849	HELD_FEM_CC	CC	CT	null	0	null	31	0	0	18	3	0	
9849	HELD_MAL_LIP	CC	CT	null	0,42	2,38	null	15	5	0	35	2	0
9883	HELD_FEM_CC	AA	AG	GG	1,64	0,46	1,55	7	9	15	1	16	5
9883	HELD_ALL_CC	AA	AG	GG	1,37	0,58	1,42	9	15	21	4	24	11
10079	CVD_ALL	AA	AG	GG	1,74	0	0,72	4	0	99	0	1	72
10079	CVD_MAL	AA	AG	GG	1,53	null	0,65	4	0	64	0	0	34
10481	HELD_FEM_ADDR5ULN	AA	AT	TT	0,4	0,85	2,53	3	6	...	8	32	33
10542	HELD_FEM_UEFF	CC	CT	TT	2,42	0,47	1,86	1	6	47	0	21	54
10542	HELD_MAL_ADDR5ULN	CC	CT	TT	null	0	null	0	0	10	0	14	55
10600	HELD_FEM_EFF	AA	AG	GG	null	0	0	0	0	21	0	4	29
10621	HELD_FEM_CC	CC	CT	TT	1,56	0,49	1,71	24	4	2	12	8	0
10745	HELD_ALL_ADDRSULN	AA	AG	GG	3,09	0,86	0,72	5	10	12	7	61	80
10745	HELD_FEM_VEFF	AA	AG	GG	0,79	1,35	0,8	11	68	74	16	45	89
10747	HELD_MAL_ADDR	CC	CT	TT	1,71	0,62	1,29	14	46	16	3	58	9
10747	CVD_ALL	CC	CT	TT	1,75	0,73	0,95	15	24	23	6	39	29
10747	HELD_MAL_ADDR3ULN	CC	CT	TT	2,24	0,45	1,77	4	16	7	3	58	9
10771	HELD_MAL_ADDRSULN	CC	CG	GG	4,67	0,67	0,42	4	4	2	6	36	28
10771	HELD_FEM_BFF	CC	CG	GG	1,14	1,07	0,86	52	118	114	40	105	131

BAYSNP	COMPARISON	GTYPE1	GTYPE2	GTYPE3	RRI	RR2	RR3	FQ1_A	FQ2_A	FQ3_A	FQ1_B	FQ2_B	FQ3_B
10870	HELD_MAL_LIP	AA	AG	GG	0	2,26	0,64	0	11	9	5	9	23
10870	HELD_FEM_LIP	AA	AG	GG	0,9	0,65	1,5	7	18	57	8	30	39
10870	HELD_MAL_CC	AA	AG	GG	0	0,52	2,51	0	3	11	2	8	8
10870	HELD_ALL_CC	AA	AG	GG	0,45	0,83	1,47	2	13	30	6	15	19
10877	HELD_ALL_HDL	AA	AC	CC	0,61	0,53	2,00	0	0	9	1	5	9
10948	HELD_FEM_LIP	GG	GT	TT	0,58	1,45	1,04	16	51	17	31	33	15
10948	HELD_ALL_LIP	GG	GT	TT	0,62	1,35	1,1	22	60	22	44	50	21
10948	HELD_FEM_CC2	GG	GT	TT	0,59	1,67	0,83	9	28	7	17	16	9
10948	CVD_MAL	GG	GT	TT	0,69	1,09	1,23	12	39	18	12	17	5
11001	HELD_MAL_DR5ULN	CC	CT	TT	5,06	1,02	0,51	2	5	3	2	37	36
11073	HELD_MAL_DR5ULN	CC	CG	GG	2,71	1,32	0,33	3	4	2	9	25	34
11153	HELD_FEM_CC	CC	CT	TT	1,76	0,57	null	24	7	0	11	11	0
11210	HELD_MAL_CC	CC	CT	TT	0,4	2,5	null	9	5	0	18	1	0
11210	HELD_ALL_DR3ULN	CC	CT	TT	0,6	1,79	0	47	16	0	125	17	2
11210	HELD_ALL_DR	CC	CT	TT	0,8	1,32	0	122	31	0	125	17	2
11248	HELD_FEM_DR	CC	CT	TT	1,57	0,59	1,08	56	19	6	38	36	5
11248	HELD_MAL_LIP	CC	CT	TT	2,65	0,38	null	15	3	0	19	15	0
11248	HELD_ALL_CC	CC	CT	TT	1,54	0,65	null	27	14	0	13	18	0
11372	HELD_MAL_LIP	AA	AG	GG	1,8	0,83	0,6	10	5	5	10	11	15
11449	HELD_FEM_CC	CC	CG	GG	1,73	0,41	2,05	1	4	26	0	10	12
11450	HELD_FEM_EFF	AA	AT	TT	1,3	1,06	0,87	28	114	147	16	107	167
11470	HELD_MAL_LIP	CC	CT	null	null	0	0	20	0	0	31	5	0

BAYSNP	COMPARISON	GTYPE1	GTYPE2	GTYPE3	RR1	RR2	RR3	FQ1_A	FQ2_A	FQ3_A	FQ1_B	FQ2_B	FQ3_B
11472	HELD_MAL_LIP	AA	AT	null	0	null	20	0	0	0	30	5	0
11472	HELD_FEM_LIP	AA	AT	null	0,61	1,63	null	75	8	0	78	2	0
11487	HELD_MAL_ADDRSULN	AT	TT	null	0	null	0	10	0	0	34	35	0
11487	HELD_MAL_ADDR3ULN	AT	TT	null	0,4	2,5	null	6	21	0	34	35	0
11488	HELD_MAL_ADDRSULN	CC	CG	null	0	0	10	0	0	0	35	32	3
11488	HELD_FEM_UEFF	CC	CG	GG	0,79	1,02	2,57	29	20	5	49	28	0
11488	HELD_MAL_ADDR3ULN	CC	CG	GG	2,48	0,3	1,52	20	4	2	35	32	3
11493	HELD_MAL_CC	AA	AG	GG	0	2,25	0,61	0	6	8	2	2	14
11502	HELD_MAL_ADDR3ULN	CC	CT	TT	0	0,69	1,94	0	8	19	7	30	36
11502	HELD_MAL_ADDRSULN	CC	CT	TT	0	0,4	3,55	0	2	8	7	30	36
11534	HELD_ALL_LIP	GG	GT	null	0	null	102	0	0	0	114	3	0
11537	CVD_FEM	AA	AG	GG	0,63	1,38	1,75	20	12	4	30	8	1
11537	HELD_FEM_EFF	AA	AG	GG	2,73	0,56	0	10	2	0	12	7	3
11560	HELD_FEM_EFF	AA	AG	GG	3	0,33	1	0	11	0	0	0	22
11578	HELD_FEM_LIP	CC	CT	null	4,62	0,22	null	60	1	0	57	8	0
11578	CVD_FEM	CC	CT	null	0,41	2,44	null	27	3	0	39	0	0
11594	HELD_FEM_ADDRSULN	CC	CT	TT	0	0	null	0	0	37	2	6	72
11594	HELD_ALL_ADDRSULN	CC	CT	TT	0	0	null	0	0	27	2	16	133
11594	HELD_ALL_CC	CC	CT	TT	null	1,6	0,62	0	10	35	0	3	38
11594	HELD_ALL adr	CC	CT	TT	0,66	0,58	1,71	1	7	147	2	16	133
11594	HELD_FEM_ADDRSULN	CC	CT	TT	0	0	null	0	0	18	2	6	72
11624	HELD_ALL_CC	CC	CT	TT	1	0,75	2,11	21	15	6	20	20	0

BAYSNP	COMPARISON	GTYPE1	GTYPE2	GTYPE3	RR1	RR2	RR3	FQ1_A	FQ2_A	FQ3_A	FQ1_B	FQ2_B	FQ3_B
11624	HELD_MAL_CC	CC	CT	TT	1,32	0,33	2,8	8	2	3	9	9	0
11624	HELD_FEM_EFF	CC	CT	TT	2,5	0,63	0	10	2	0	12	6	3
11627	HELD_ALL_CC	CC	CT	TT	0,86	0,86	2,05	20	18	7	21	19	0
11627	HELD_MAL_CC	CC	CT	TT	1	0,58	2,64	7	4	3	9	9	0
11627	HELD_FEM_EFF	CC	CT	TT	2,73	0,56	0	10	2	0	12	7	3
11644	HELD_MAL_ADDRSULN	AA	AG	GG	0	0,45	3,26	0	2	8	7	26	35
11650	HELD_FEM_EFF	AA	AG	GG	1,07	0,8	1,21	26	105	160	23	135	132
11654	HELD_ALL_ADDRSULN	AA	AG	GG	2,59	0,24	1,48	7	3	15	14	56	66
11654	HELD_FEM_ADDRSULN	AA	AG	GG	2,81	0,12	1,65	5	1	9	8	31	32
11654	HELD_FEM_ADDR3ULN	AA	AG	GG	1,81	0,48	1,25	8	7	17	8	31	32
11654	HELD_ALL_ADDR3ULN	AA	AG	GG	1,83	0,66	1,02	12	15	26	14	56	66
11655	HELD_ALL_ADDRSULN	AA	AC	CC	1,56	0,24	2,3	16	3	7	72	59	17
11655	HELD_FEM_ADDRSULN	AA	AC	CC	2,03	0,11	2,11	11	1	5	35	34	11
11655	HELD_FEM_ADDR3ULN	AA	AC	CC	1,34	0,45	1,64	19	7	9	35	34	11
11656	HELD_MAL_LIP	CC	CT	TT	0,53	0,96	2,57	6	8	6	19	15	2
11656	HELD_FEM_EFF	CC	CT	TT	2,57	0,56	0	7	5	0	5	14	3
11656	HELD_ALL_LIP	CC	CT	TT	0,79	1,01	1,5	35	49	18	51	54	9
11825	HELD_MAL_ADDRSULN	AA	AG	null	0,25	4	null	6	3	0	58	5	0
11914	HELD_MAL_ADDRSULN	AA	AT	TT	0,11	0	9,83	1	0	8	41	1	27
11914	HELD_ALL_ADDRSULN	AA	AT	TT	0,45	1,43	1,48	6	12	9	63	52	36
12008	HELD_FEM_EFF	CC	CT	null	0,72	1,38	null	251	27	0	264	13	0
12008	HELD_ALL_ADDRSULN	CC	CT	null	null	0	null	24	0	0	122	12	0

BAYSNP	COMPARISON	GTYPE1	GTYPE2	GTYPE3	RRJ	RR2	RR3	FQ1_A	FQ2_A	FQ3_A	FQ1_B	FQ2_B	FQ3_B
12097	HELD_ALL_ADRSULN	AG	GG	null	2,66	0,38	null	6	22	0	11	144	0
12097	HELD_FEM_ADR3ULN	AG	GG	null	2,05	0,49	null	7	31	0	5	78	0
12097	HELD_MAL_ADR5ULN	AG	GG	null	3,48	0,29	null	3	7	0	6	66	0
12097	HELD_ALL_ADR3ULN	AG	GG	null	1,77	0,56	null	10	53	0	11	144	0
12366	HELD_FEM_UEFF	AA	AG	GG	1,33	1,02	0	32	18	0	39	26	9
12366	HELD_ALL_ADRSULN	AA	AG	GG	1,82	0,34	2,26	18	4	3	85	59	7
12619	HELD_MAL_ADR5ULN	AG	GG	null	8,89	0,11	null	1	9	0	0	71	0
12619	HELD_ALL_ADRSULN	AG	GG	null	4,67	0,21	null	2	25	0	1	150	0
13025	HELD_ALL_ADR5ULN	AA	AC	CC	1,12	0,51	2,38	13	8	7	65	71	15
13191	HELD_FEM_LPP	AA	AG	GG	0,71	0,71	1,55	6	30	47	10	42	27
13191	HELD_MAL_CC	AA	AG	GG	2,5	1,67	0,43	2	7	5	0	5	13
13191	HELD_ALL_LPP	AA	AG	GG	0,65	0,81	1,38	6	39	56	13	55	46
13937	HELD_FEM_ADRSULN	AA	AC	CC	0,36	1,91	2,53	4	11	2	42	38	3
900002	CVD_FEM	GG	GT	TT	1,65	1,29	0,64	5	13	16	2	11	27
900013	CVD_FEM	CC	CG	GG	1,7	0,47	1,34	20	9	6	13	23	4
900013	CVD_ALL	CC	CG	GG	1,32	0,7	1,16	58	34	12	29	39	6
900025	CVD_MAL	GG	GT	TT	0,73	0,88	1,3	7	27	32	7	17	10
900032	CVD_FEM	CC	CT	TT	2,48	0,22	2,54	23	1	1	28	9	0
900045	HELD_FEM_BFF	CC	CT	TT	0,42	0,48	2,67	1	2	9	5	8	9
900065	CVD_FEM	AA	AC	CC	1,91	0,7	0	22	10	0	16	18	5
900065	CVD_MAL	AA	AC	CC	1,29	0,72	1,53	25	30	4	7	22	0
900065	CVD_ALL	AA	AC	CC	1,36	0,77	0,77	47	40	4	23	40	5

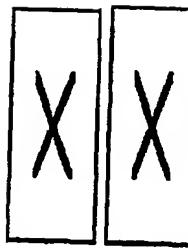
BAYSNP	COMPARISON	GTYPE1	GTYPE2	GTYPE3	RR1	RR2	RR3	FQ1_A	FQ2_A	FQ3_A	FQ1_B	FQ2_B	FQ3_B
900078	HELD_ALL_ADR3ULN	AA	AG	GG	0,56	1,79	null	52	12	0	142	13	0
900078	HELD_ALL_ADR5ULN	AA	AG	GG	0,41	2,45	null	21	6	0	142	13	0
900078	HELD_FEM_ADR3ULN	AA	AG	GG	0,49	2,05	null	31	7	0	78	5	0
900082	HELD_FEM_ADR3ULN	AA	AG	GG	1	0,49	1,9	8	9	18	17	36	21
900082	HELD_FEM_ADR5ULN	AA	AG	GG	0,76	0,39	2,76	3	4	10	17	36	21
900096	CVD_ALL	AA	AG	GG	0,74	1,35	1,15	60	37	4	55	15	2
900107	HELD_MAL_ADR5ULN	CC	CT	TT	0	0,52	3,06	0	2	8	9	25	39
900115	HELD_MAL_ADR5ULN	AA	AG	GG	0,24	0,78	4,6	1	4	4	27	37	8
900115	HELD_FEM_EFF	AA	AG	GG	1,47	0,56	1,8	22	14	4	17	28	1
900121	HELD_MAL_ADR	GG	GT	TT	0,46	1,42	0,95	5	37	24	15	26	26
900173	CVD_ALL	GG	GT	TT	0,5	1,35	1,38	5	7	11	11	4	7
10000002	HELD_FEM_EFF	AA	AG	GG	2,67	0,8	0	9	3	0	9	7	6
10000006	HELD_FEM_CC	AA	AG	GG	3,35	0,26	0,56	28	2	1	11	9	2
10000006	HELD_ALL_CC	AA	AG	GG	2,52	0,41	0,45	39	4	1	23	12	3
10000014	HELD_ALL_CC	AA	AC	CC	2,18	0,33	1,26	40	3	2	26	12	1
10000014	HELD_FEM_CC	AA	AC	CC	2,17	0,34	1,73	28	2	1	15	7	0
10000025	HELD_MAL_LP	CC	CT	TT	1,17	1,41	0	9	11	0	14	15	7

Table 6b: Correlation of PA SNP alleles to relative risk

For diagnostic conclusions to be drawn from genotyping a particular patient we calculated the relative risks RR1, and RR2 for the two possible alleles of each SNP. Given the allele frequencies as

	allele1	allele2
case	N11	N12
control	N21	N22

we calculate



Here, the *case* and *control* populations represent any case-control-group pair, or bad(case)-good(control)-group pair, respectively (due to their increased response to statins, 'high responders' are treated as a case cohort, whereas 'low responders' are treated as the respective control cohort). A value $RR1 > 1$, and $RR2 > 1$ indicates an increased risk for individuals carrying allele 1, and allele2, respectively. For example, $RR1=3$ indicates a 3-fold risk of an individual carrying allele 1 as compared to individuals not carrying allele 1 (a detailed description of relative risk calculation and statistics can be found in (Biostatistics, L. D. Fisher and G. van Belle, Wiley Interscience 1993)). The baySNP number refers to an internal numbering of the PA SNPs and can be found in the sequence listing. null: not defined.

BAYSNP	ALLELE1	ALLELE2	COMPARISON	RR1	RR2	SIZE A	FREQ1 A	FREQ2 A	SIZE B	FREQ1 B	FREQ2 B
28	C	T	HELD_FEM_EFF	0,42	2,39	12	4	20	22	18	26
29	A	G	HELD_ALL_HDL	2,01	0,5	10	12	8	15	7	23
29	A	G	HELD_MAL_ADR3ULN	1,63	0,61	26	33	19	72	68	76
29	A	G	HELD_MAL_ADR5ULN	2,6	0,38	9	13	5	72	68	76
52	C	G	HELD_FEM_EFF	1,84	0,54	18	24	12	31	27	35
56	A	G	HELD_FEM_EFF	2,29	0,44	12	5	19	22	2	42
89	A	G	HELD_ALL_CC	null	0	45	90	0	40	77	3
90	C	T	HELD_FEM_CC	0,78	1,27	31	29	33	22	27	17
99	C	T	HELD_FEM_LIP	1,02	0,98	82	54	110	80	51	109
140	C	T	HELD_FEM_EFF	null	0	12	24	0	21	4	38
152	A	G	HELD_FEM_EFF	0,51	1,96	12	12	12	22	33	11
214	A	G	HELD_ALL_LIP	1	1	97	156	38	113	182	44
214	A	G	HELD_FEM_LIP	1,09	0,92	81	131	31	78	122	34
221	C	G	HELD_ALL_CC	0,88	1,13	45	26	64	39	27	51
221	C	G	HELD_FEM_CC	0,77	1,3	31	17	45	22	18	26
224	C	T	HELD_FEM_LIP	0,79	1,27	79	110	48	79	125	33
224	C	T	HELD_MAL_LIP	2,28	0,44	20	35	5	37	51	23
294	C	T	HELD_ALL_CC	0,81	1,24	45	56	34	40	58	22
307	C	T	CVD_FEM	0,57	1,75	36	19	53	38	38	38
307	C	T	HELD_ALL_LIP	1,2	0,83	102	70	134	117	63	171
411	A	T	HELD_ALL_HDL	1,56	0,64	10	17	3	15	18	12
449	C	G	HELD_MAL_LIP	0,41	2,47	20	3	37	37	16	58
466	C	T	CVD_FEM	0,7	1,43	35	27	43	40	44	36

BAYSNP	ALLELE1	ALLELE2	COMPARISON	RR1	RR2	SIZE A	FREQ1 A	FREQ2 A	SIZE B	FREQ1 B	FREQ2 B
472	A	G	HELD_FEM_EFF	null	0	11	22	0	22	12	32
542	A	G	HELD_MAL_CC	2,79	0,36	14	12	16	19	2	36
542	A	G	HELD_MAL_HDL	3,66	0,27	21	14	28	27	3	51
542	A	G	HELD_ALL_ADR	1,19	0,84	159	53	265	154	37	271
542	A	G	HELD_FEM_HDL	0,66	1,51	23	2	44	32	10	54
739	C	G	HELD_ALL_CC	0,73	1,37	45	39	51	40	48	32
821	A	C	HELD_MAL_LIP2	1,12	0,9	309	180	438	349	174	524
821	A	C	HELD_FEM_VEFF	0,42	2,4	10	4	16	14	14	14
1005	A	G	HELD_MAL_CC	2,7	0,37	14	26	2	18	27	9
1055	A	T	HELD_MAL_CC	0,56	1,77	9	3	15	12	8	16
1056	A	G	HELD_FEM_EFF	1,01	0,99	24	30	18	33	41	25
1085	A	G	HELD_MAL_LIP	0,57	1,74	20	17	23	36	46	26
1085	A	G	CVD_FEM	1,53	0,65	34	51	17	40	47	33
1086	A	G	HELD_MAL_LIP	1,73	0,58	20	24	16	36	28	44
1092	C	G	HELD_MAL_LIP	0,58	1,72	20	9	31	37	29	45
1096	G	T	HELD_MAL_CC	1,8	0,56	14	7	21	18	3	33
1096	G	T	CVD_MAL	0,93	1,08	69	21	117	33	12	54
1101	C	T	HELD_FEM_EFF	null	0	12	24	0	22	40	4
1204	A	G	HELD_MAL_LIP	1,91	0,52	19	12	26	35	9	61
1204	A	G	HELD_ALL_LIP	1,26	0,8	99	62	136	115	52	178
1504	C	T	HELD_ALL_CC	0,92	1,08	44	37	51	39	36	42
1504	C	T	HELD_MAL_LIP	0,69	1,46	19	12	26	37	33	41
1504	C	T	HELD_MAL_CC	1,35	0,74	14	13	15	18	12	24

BAYSNP	ALLELE1	ALLELE2	COMPARISON	RR1	RR2	SIZE A	FREQ1 A	FREQ2 A	SIZE B	FREQ1 B	FREQ2 B
1504	C	T	HELD_FEM_CC	0,75	1,33	30	24	36	21	24	18
1511	G	T	HELD_FEM_EFF	0,6	1,67	12	15	9	22	35	9
1524	A	C	HELD_FEM_DR3ULN	0,9	1,11	38	16	60	82	39	125
1556	C	G	HELD_FEM_EFF	2,39	0,42	12	7	17	22	3	41
1561	A	C	CVD_FEM	1,53	0,65	36	58	14	40	53	27
1582	C	T	HELD_MAL_LIP	0,46	2,17	20	5	35	37	22	52
1638	A	G	HELD_FEM_CC	0,62	1,6	31	10	52	22	15	29
1653	G	T	CVD_MAL	1,07	0,93	69	70	68	33	30	36
1662	C	T	HELD_MAL_CC	0,18	5,5	14	8	20	18	36	0
1714	A	G	CVD_MAL	0,78	1,28	66	32	100	34	26	42
1722	C	T	HELD_FEM_DR5ULN	1,61	0,62	18	21	15	81	71	91
1757	A	G	HELD_FEM_EFF	1,41	0,71	20	15	25	32	16	48
1765	A	G	HELD_ALL_DR3ULN	0,42	2,35	63	9	117	149	56	242
1765	A	G	HELD_ALL_DR3ULN	0,42	2,35	63	9	117	149	56	242
1765	A	G	HELD_ALL_DR5ULN	0,29	3,42	27	3	51	149	56	242
1765	A	G	HELD_ALL_DR5ULN	0,29	3,42	27	3	51	149	56	242
1765	A	G	HELD_MAL_DR3ULN	0,24	4,09	26	2	50	70	25	115
1765	A	G	HELD_MAL_DR3ULN	0,24	4,09	26	2	50	70	25	115
1765	A	G	HELD_MAL_DR5ULN	null	0	10	20	0	70	25	115
1765	A	G	HELD_MAL_DR5ULN	null	0	10	20	0	70	25	115
1765	A	G	HELD_FEM_DR3ULN	0,53	1,87	37	7	67	79	31	127
1765	A	G	HELD_FEM_DR3ULN	0,53	1,87	37	7	67	79	31	127
1776	A	G	HELD_ALL_CC	null	0	45	90	0	40	74	6

BAYSNP	ALLELE1	ALLELE2	COMPARISON	RRI	SIZE A	FREQ1 A	FREQ2 A	SIZE B	FREQ1 B	FREQ2 B
1776	A	G	HELD_FEM_CC	null	0	31	62	0	22	40
1799	C	T	HELD_FEM_LIP2	0,93	1,07	291	365	217	356	468
1799	C	T	HELD_MAL_CC	0,56	1,77	14	15	13	18	28
1806	A	G	HELD_FEM_EFF	4,44	0,23	12	23	1	22	34
1837	C	T	HELD_FEM_LIP2	1,04	0,96	304	436	172	355	499
1837	C	T	HELD_ALL_LIP2	1,1	0,91	607	891	323	682	952
1837	C	T	HELD_ALL_ADDSULN	2,03	0,49	28	46	10	155	208
1837	C	T	HELD_MAL_ADR	1,24	0,81	77	107	47	72	86
1837	C	T	HELD_MAL_LIP2	1,17	0,86	303	455	151	327	453
1870	C	T	HELD_ALL_CC	1,3	0,77	45	29	61	39	16
1870	C	T	HELD_FEM_CC	1,33	0,75	31	22	40	22	9
1882	C	T	CVD_MAL	0,92	1,08	69	79	59	34	43
1988	C	T	HELD_ALL_LIP	1,27	0,79	100	143	57	116	144
2000	C	T	CVD_MAL	2,45	0,41	70	136	4	34	58
2000	C	T	CVD_ALL	1,98	0,51	105	202	8	74	130
2000	C	T	HELD_FEM_CC2	3,29	0,3	46	90	2	42	74
2000	C	T	HELD_MAL_HDL	2	0,5	20	40	0	22	40
2000	C	T	HELD_FEM_ADR	2,01	0,5	79	154	4	82	152
2000	C	T	HELD_MAL_CC	0,51	1,98	14	22	6	19	36
2071	A	G	CVD_ALL	1,22	0,82	102	80	124	74	42
2078	G	T	HELD_MAL_LIP	1,74	0,58	18	13	23	35	13
2085	G	T	HELD_FEM_VEFF	2,62	0,38	10	16	4	14	13
2095	A	G	CVD_ALL	0,03	37,5	105	4	206	73	146

BAYSNP	ALLELE1	ALLELE2	COMPARISON	RR1	RR2	SIZE A	FREQ1 A	FREQ2 A	SIZE B	FREQ1 B	FREQ2 B
2119	A	G	HELD_MAL_LIP	0,68	1,48	20	23	17	37	53	21
2119	A	G	HELD_ALL_LIP	0,85	1,17	102	131	73	117	166	68
2119	A	G	HELD_FEM_EFF	0,6	1,67	12	15	9	22	35	9
2141	A	G	HELD_FEM_EFF	1,56	0,64	12	6	18	22	6	38
2141	A	G	HELD_ALL_CC	0,99	1,01	45	17	73	39	15	63
2182	A	G	HELD_FEM_EFF	2,82	0,35	12	18	6	21	16	26
2234	G	T	HELD_MAL_LIP	0,54	1,85	20	10	30	35	32	38
2281	A	C	HELD_FEM_VEFF	0,46	2,17	9	5	13	13	15	11
2298	A	C	CVD_FEM	0,98	1,02	35	18	52	38	20	56
2298	A	C	HELD_MAL_CC2	0,6	1,67	29	8	50	28	16	40
2341	C	T	HELD_FEM_CC	0,12	8,33	31	6	56	22	44	0
2357	A	G	HELD_ALL_CC2	1,04	0,96	74	28	120	71	25	117
2357	A	G	HELD_ALL_CC	1,01	0,99	45	16	74	40	14	66
2357	A	G	HELD_MAL_LIP	0,48	2,08	20	4	36	36	17	55
2357	A	G	HELD_FEM_CC	1,1	0,91	31	12	50	22	7	37
2366	G	T	CVD_FEM	1,51	0,66	33	38	28	40	31	49
2423	A	G	CVD_FEM	1,57	0,63	33	45	21	39	38	40
2708	C	T	CVD_FEM	3,51	0,29	29	57	1	40	73	7
2995	A	C	HELD_FEM_ADDRESSLN	1,82	0,55	18	16	20	82	45	119
2995	A	C	HELD_FEM_UEFF	0,71	1,41	54	24	84	75	50	100
3360	G	T	HELD_MAL_ADDRESSLN	null	0	10	20	0	73	122	24
3464	A	G	HELD_ALL_CC	0,62	1,61	45	21	69	40	35	45
3464	A	G	HELD_FEM_CC	0,61	1,63	31	13	49	22	19	25

BAYSNP	ALLELE1	ALLELE2	COMPARISON	RRI	RR2	SIZEA	FREQ1A	FREQ2A	SIZEB	FREQ1B	FREQ2B
3689	C	G	HELD_FEM_EFF	3,32	0,3	6	9	3	14	10	18
3975	A	C	HELD_FEM_UEFF	0,68	1,47	56	28	84	75	58	92
3976	A	G	HELD_FEM_UEFF	0,69	1,44	56	28	84	75	57	93
4206	A	T	HELD_FEM_ADDR3ULN	0,69	1,45	37	36	38	83	103	63
4838	A	G	HELD_FEM_VEFF	2,4	0,42	10	16	4	14	14	14
4838	A	G	HELD_FEM_VEFF	2,4	0,42	10	16	4	14	14	14
4912	A	G	HELD_FEM_VEFF	2,05	0,49	12	14	10	20	12	28
4925	A	C	HELD_MAL_CC	0,56	1,8	14	21	7	18	33	3
4966	A	G	HELD_MAL_ADDR3ULN	0,72	1,39	26	22	30	72	77	67
5014	A	G	HELD_ALL_ADDR5ULN	0,54	1,85	28	8	48	152	77	227
5014	A	G	HELD_FEM_ADDR5ULN	0,6	1,67	18	5	31	81	37...	125
5296	A	G	CVD_FEM	1,59	0,63	36	10	62	40	4	76
5296	A	G	HELD_FEM_EFF	0,67	1,5	12	3	21	22	9	35
5296	A	G	CVD_ALL	1,29	0,78	104	27	181	74	10	138
5298	C	T	HELD_FEM_EFF	0,71	1,41	11	3	19	22	9	35
5298	C	T	CVD_ALL	1,32	0,76	101	28	174	74	10	138
5298	C	T	CVD_FEM	1,62	0,62	35	10	60	40	4	76
5320	A	G	HELD_FEM_EFF	0,52	1,93	19	12	26	33	37	29
5361	A	C	CVD_MAL	0,82	1,22	64	53	75	32	36	28
5457	A	G	HELD_FEM_EFF	0,51	1,96	12	2	22	21	8	34
5704	C	T	HELD_MAL_LIP	0,57	1,75	20	10	30	37	32	42
5704	C	T	CVD_MAL	0,79	1,27	68	40	96	33	30	36

BAYSNP	ALLELE1	ALLELE2	COMPARISON	RR1	RR2	SIZE A	FREQ1 A	FREQ2 A	SIZE B	FREQ1 B	FREQ2 B
5717	A	G	HELD_FEM_ADR3ULN	1,58	0,63	38	50	26	83	83	83
5717	A	G	HELD_ALL_ADR3ULN	1,36	0,74	65	74	56	156	144	168
5959	A	G	HELD_ALL_CC	1,53	0,65	43	52	34	38	29	47
5959	A	G	CVD_FEM	2,63	0,38	9	12	6	13	7	19
5959	A	G	HELD_MAL_CC	1,71	0,59	14	15	13	17	10	24
5959	A	G	HELD_MAL_ADR5ULN	0,54	1,85	9	6	12	67	67	67
5959	A	G	HELD_FEM_ADR	1,26	0,79	72	71	73	68	51	85
6162	C	G	HELD_ALL_ADR3ULN	0,97	1,03	64	37	91	151	90	212
6162	C	G	HELD_ALL_ADR	0,96	1,04	156	88	224	151	90	212
6162	C	G	HELD_ALL_ADR5ULN	0,99	1,01	27	16	38	151	90	212
6162	C	G	HELD_MAL_ADR3ULN	0,82	1,22	26	13	39	71	43	99
6162	C	G	HELD_FEM_ADR5ULN	1,28	0,78	18	13	23	80	47	113
6162	C	G	HELD_MAL_ADR	0,92	1,08	74	40	108	71	43	99
6236	C	T	HELD_ALL_ADR5ULN	1,85	0,54	27	24	30	152	84	220
6236	C	T	HELD_MAL_ADR3ULN	1,67	0,6	27	23	31	72	38	106
6236	C	T	HELD_MAL_ADR5ULN	2,42	0,41	10	10	72	38	106	
6236	C	T	HELD_ALL_ADR3ULN	1,36	0,74	63	47	79	152	84	220
6482	A	G	HELD_MAL_HDL	0,51	1,96	17	18	16	21	34	8
6482	A	G	HELD_ALL_LIP2	0,91	1,1	619	918	320	709	1098	320
6482	A	G	HELD_MAL_CC2	1,82	0,55	27	43	11	28	32	24
6482	A	G	HELD_MAL_LIP2	0,87	1,15	309	461	157	339	539	139
6498	A	G	CVD_FEM	2,18	0,46	32	60	4	35	57	13
6744	C	T	HELD_ALL_ADR5ULN	1,82	0,55	26	21	31	149	74	224

BAYSNP'	ALLELE1	ALLELE2	COMPARISON	RRI	RR2	SIZE A	FREQ1A	SIZE B	FREQ1B	FREQ2A	FREQ2B
7133	C	G	HELD_MAL_CC	0,36	2,8	14	20	8	18	36	0
8021	A	G	CVD_FEM	1,03	0,97	28	35	21	36	44	28
8060	A	G	CVD_FEM	1,66	0,6	35	65	5	40	68	12
8060	A	G	HELD_FEM_HDL	0,5	1,99	18	29	7	23	43	3
8210	A	G	HELD_FEM_EFF	0,72	1,4	12	9	15	22	22	22
8592	C	T	HELD_FEM_VEFF	0,99	1,01	150	122	178	143	118	168
8816	C	G	HELD_FEM_EFF	1,91	0,52	13	15	11	11	5	17
8846	A	G	HELD_ALL_LIP	1,11	0,9	107	161	53	116	166	66
8943	A	C	HELD_MAL_LIP	2,17	0,46	20	35	5	37	52	22
9193	C	G	HELD_FEM_LIP	1,48	0,68	83	155	11	80	140	20
9193	C	G	CVD_FEM	0,6	1,67	36	63	9	40	77	3
9443	C	T	CVD_MAL	1,23	0,82	69	43	95	33	12	54
9516	A	G	HELD_MAL_CC	1,87	0,54	14	17	11	18	12	24
9698	A	G	HELD_MAL_ADR	0,38	2,62	74	8	140	72	30	114
9698	A	G	HELD_MAL_ADDR3ULN	null	0	27	54	0	72	30	114
9698	A	G	HELD_FEM_EFF	0,91	1,1	294	105	483	298	123	473
9698	A	G	HELD_MAL_ADDR5ULN	0	10	20	0	72	30	114	
9698	A	G	CVD_ALL	1,27	0,79	102	46	158	72	19	125
9849	C	T	HELD_FEM_CC	null	0	31	62	0	21	39	3
9849	C	T	HELD_MAL_LIP	0,46	2,18	20	35	5	37	72	2
9883	A	G	HELD_FEM_CC	0,93	1,07	31	23	39	22	18	26
9883	A	G	HELD_ALL_CC	0,92	1,09	45	33	57	39	32	46
10079	A	G	CVD_ALL	1,54	0,65	103	8	198	73	1	145

BAYSNP	ALLELE1	ALLELE2	COMPARISON	RR1	RR2	SIZE A	FREQ1 A	FREQ2 A	SIZE B	FREQ1 B	FREQ2 B
10079	A	G	CVD_MAL	0,11	9,5	68	8	128	34	68	0
10481	A	T	HELD_FEM_ADDRESSLN	0,46	2,2	17	12	22	83	97	69
10542	C	T	HELD_FEM_UEFF	0,63	1,58	54	8	100	75	21	129
10542	C	T	HELD_MAL_ADDRESSLN	null	0	10	20	0	69	14	124
10600	A	G	HELD_FEM_EFF	null	0	21	42	0	33	4	62
10621	C	T	HELD_FEM_CC	1,24	0,81	30	52	8	20	32	8
10745	A	G	HELD_ALL_ADDRESSLN	1,58	0,63	27	20	34	148	75	221
10745	A	G	HELD_FEM_VEFF	1,1	0,91	153	90	216	150	77	223
10747	C	T	HELD_MAL adr	1,06	0,94	76	74	78	70	64	76
10747	C	T	CVD_ALL	1,23	0,82	62	54	70	74	51	97
10747	C	T	HELD_MAL_ADDRESSLN	0,96	1,04	27	24	30	70	64	76
10771	C	G	HELD_MAL_ADDRESSLN	2,5	0,4	10	12	8	70	48	92
10771	C	G	HELD_FEM_EFF	1,12	0,89	284	222	346	276	185	367
10870	A	G	HELD_MAL_LIP	1,06	0,94	20	11	29	37	19	55
10870	A	G	HELD_FEM_LIP	0,75	1,34	82	32	132	77	46	108
10870	A	G	HELD_MAL_CC	0,39	2,55	14	3	25	18	12	24
10870	A	G	HELD_ALL_CC	0,67	1,5	45	17	73	40	27	53
10877	A	C	HELD_ALL_HDL	3,57	0,28	9	18	0	15	7	23
10948	G	T	HELD_FEM_LIP	0,81	1,23	84	83	85	79	95	63
10948	G	T	HELD_ALL_LIP	0,81	1,23	104	104	104	115	138	92
10948	G	T	CVD_FEM_CC2	0,87	1,15	44	46	42	42	50	34
10948	G	T	CVD_MAL	0,82	1,21	69	63	75	34	41	27
11001	C	T	HELD_MAL_ADDRESSLN	1,96	0,51	10	9	11	75	41	109

BAYSNP	ALLELE1	ALLELE2	COMPARISON	RR1	RR2	SIZEA	FREQ1A	FREQ2A	SIZEB	FREQ1B	FREQ2B
11073	C	G	HELD_MAL_ADR5ULN	2,38	0,42	9	10	8	68	43	93
11153	C	T	HELD_FEM_CC	1,61	0,62	31	55	7	22	33	11
11210	C	T	HELD_MAL_CC	0,46	2,17	14	23	5	19	37	1
11210	C	T	HELD_ALL_ADR3ULN	0,67	1,48	63	110	16	144	267	21
11210	C	T	HELD_ALL_ADR	0,85	1,17	153	275	31	144	267	21
11248	C	T	HELD_FEM_ADR	1,34	0,75	81	131	31	79	112	46
11248	C	T	HELD_MAL_LIP	2,3	0,43	18	33	3	34	53	15
11248	C	T	HELD_ALL_CC	1,39	0,72	41	68	14	31	44	18
11372	A	G	HELD_MAL_LIP	1,67	0,6	20	25	15	36	31	41
11449	C	G	HELD_FEM_CC	0,6	1,66	31	6	56	22	10	34
11450	A	T	HELD_FEM_EFF	1,14	0,87	289	170	408	290	139	441
11470	C	T	HELD_MAL_LIP	null	0	20	40	0	36	67	5
11472	A	T	HELD_MAL_LIP	null	0	20	40	0	35	65	5
11472	A	T	HELD_FEM_LIP	0,63	1,6	83	158	8	80	158	2
11487	A	T	HELD_MAL_ADR5ULN	null	0	10	20	0	69	34	104
11487	A	T	HELD_MAL_ADR3ULN	0,48	2,11	27	6	48	69	34	104
11488	C	G	HELD_MAL_ADR5ULN	null	0	10	20	0	70	102	38
11488	C	G	HELD_FEM_UEFF	0,74	1,35	54	78	30	77	126	28
11488	C	G	HELD_MAL_ADR3ULN	1,73	0,58	26	44	8	70	102	38
11493	A	G	HELD_MAL_CC	1,18	0,85	14	6	22	18	6	30
11502	C	T	HELD_MAL_ADR3ULN	0,49	2,02	27	8	46	73	44	102
11502	C	T	HELD_MAL_ADR5ULN	0,29	3,45	10	2	18	73	44	102
11534	G	T	HELD_ALL_LIP	null	0	102	204	0	117	231	3

BAYSNP	ALLELE1	ALLELE2	COMPARISON	RR1	RR2	SIZE_A	FREQ1_A	FREQ2_A	SIZE_B	FREQ1_B	FREQ2_B
11537	A	G	CVD_FEM	0,65	1,54	36	52	20	39	68	10
11537	A	G	HELD_FEM_EFF	3,11	0,32	12	22	2	22	31	13
11560	A	G	HELD_FEM_EFF	0,04	23	12	2	22	22	44	0
11578	C	T	HELD_FEM_LIP	4,48	0,22	61	121	1	65	122	8
11578	C	T	CVD_FEM	0,42	2,37	30	57	3	39	78	0
11594	C	T	HELD_FEM_ADDR3ULN	null	0	37	74	0	80	10	150
11594	C	T	HELD_ALL_ADDRSULN	null	0	27	54	0	151	20	282
11594	C	T	HELD_ALL_CC	1,53	0,65	45	10	80	41	3	79
11594	C	T	HELD_ALL adr	0,6	1,66	155	9	301	151	20	282
11594	C	T	HELD_FEM_ADDRSULN	null	0	18	36	0	80	10	150
11624	C	T	HELD_ALL_CC	0,85	1,18	42	57	27	40	60	20
11624	C	T	HELD_MAL_CC	0,85	1,18	13	18	8	18	27	9
11624	C	T	HELD_FEM_EFF	2,96	0,34	12	22	2	21	30	12
11627	C	T	HELD_ALL_CC	0,78	1,29	45	58	32	40	61	19
11627	C	T	HELD_MAL_CC	0,76	1,32	14	18	10	18	27	9
11627	C	T	HELD_FEM_EFF	3,11	0,32	12	22	2	22	31	13
11644	A	G	HELD_MAL_ADDRSULN	0,3	3,32	10	2	18	68	40	96
11650	A	G	HELD_FEM_EFF	0,9	1,11	291	157	425	290	181	399
11654	A	G	HELD_ALL_ADDRSULN	1,13	0,89	25	17	33	136	84	188
11654	A	G	HELD_FEM_ADDRSULN	1,14	0,88	15	11	19	71	47	95
11654	A	G	HELD_FEM_ADDR3ULN	1,09	0,92	32	23	41	71	47	95
11654	A	G	HELD_ALL_ADDR3ULN	1,21	0,83	53	39	67	136	84	188
11655	A	C	HELD_ALL_ADDRSULN	0,95	1,05	26	35	17	148	203	93

BAYSNP	ALLELE1	ALLELE2	COMPARISON	RR1	RR2	SIZE A	FREQ1 A	FREQ2 A	SIZE B	FREQ1 B	FREQ2 B
11655	A	C	HELD_FEM_ADR5ULN	1,1	0,91	17	23	11	80	104	56
11655	A	C	HELD_FEM_ADR3ULN	0,98	1,02	35	45	25	80	104	56
11656	C	T	HELD_MAL_LIP	0,53	1,87	20	20	20	36	53	19
11656	C	T	HELD_FEM_EFF	2,21	0,45	12	19	5	22	24	20
11656	C	T	HELD_ALI_LIP	0,8	1,25	102	119	85	114	156	72
11825	A	G	HELD_MAL_ADR5ULN	0,29	3,4	9	15	3	63	121	5
11914	A	T	HELD_MAL_ADR5ULN	0,1	9,58	9	2	16	69	83	55
11914	A	T	HELD_ALL_ADR5ULN	0,61	1,64	27	24	30	151	178	124
12008	C	T	HELD_FEM_EFF	0,73	1,37	278	529	27	277	541	13
12008	C	T	HELD_ALL_ADR5ULN	null	0	24	48	0	134	256	12
12097	A	G	HELD_ALL_ADR5ULN	2,46	0,41	28	6	50	155	11	299
12097	A	G	HELD_FEM_ADR3ULN	1,94	0,51	38	7	69	83	5	161
12097	A	G	HELD_MAL_ADR5ULN	3,04	0,33	10	3	17	72	6	138
12097	A	G	HELD_ALL_ADR3ULN	1,7	0,59	63	10	116	155	11	299
12366	A	G	HELD_FEM_UEFF	1,52	0,66	50	82	18	74	104	44
12366	A	G	HELD_ALL_ADR5ULN	1,23	0,81	25	40	10	151	229	73
12619	A	G	HELD_MAL_ADR5ULN	0,01	143	10	1	19	71	142	0
12619	A	G	HELD_ALL_ADR5ULN	4,53	0,22	27	2	52	151	1	301
13025	A	C	HELD_ALL_ADR5ULN	0,81	1,24	28	34	22	151	201	101
13191	A	G	HELD_FEM_LIP	0,72	1,4	83	42	124	79	62	96
13191	A	G	HELD_MAL_CC	1,94	0,52	14	11	17	18	5	31
13191	A	G	HELD_ALL_LIP	0,76	1,31	101	51	151	114	81	147
13937	A	C	HELD_FEM_ADR5ULN	0,53	1,89	17	19	15	83	122	44

BAYSNP	ALLELE1	ALLELE2	COMPARISON		RR1	RR2	SIZE A	FREQ1 A	FREQ2 A	SIZE B	FREQ1 B	FREQ2 B
			CVD_FEM	CVD_MAL								
900002	G	T	CVD_FEM	1,48	0,68	34	23	45	40	15	65	
900013	C	G	CVD_FEM	1,24	0,81	35	49	21	40	49	31	
900013	C	G	CVD_ALL	1,14	0,88	104	150	58	74	97	51	
900025	G	T	CVD_MAL	0,8	1,25	66	41	91	34	31	37	
900032	C	T	CVD_FEM	1,68	0,6	25	47	3	37	65	9	
900045	C	T	HELD_FEM_EFF	0,42	2,39	12	4	20	22	18	26	
900065	A	C	CVD_FEM	1,97	0,51	32	54	10	39	50	28	
900065	A	C	CVD_MAL	1,09	0,92	59	80	38	29	36	22	
900065	A	C	CVD_ALL	1,24	0,8	91	134	48	68	86	50	
900078	A	G	HELD_ALL_ADR3ULN	0,59	1,71	64	116	12	155	297	13	
900078	A	G	HELD_ALL_ADR5ULN	0,44	2,27	27	48	6	155	297	13	
900078	A	G	HELD_FEM_ADR3ULN	0,51	1,94	38	69	7	83	161	5	
900082	A	G	HELD_FEM_ADR3ULN	0,72	1,39	35	25	45	74	70	78	
900082	A	G	HELD_FEM_ADR5ULN	0,53	1,88	17	10	24	74	70	78	
900096	A	G	CVD_ALL	0,79	1,26	101	157	45	72	125	19	
900107	C	T	HELD_MAL_ADR5ULN	0,3	3,35	10	2	18	73	43	103	
900115	A	G	HELD_MAL_ADR5ULN	0,34	2,98	9	6	12	72	91	53	
900115	A	G	HELD_FEM_EFF	1,14	0,88	40	58	22	46	62	30	
900121	G	T	HELD_MAL_ADR	0,88	1,14	66	47	85	67	56	78	
900173	G	T	CVD_ALL	0,64	1,56	23	17	29	22	26	18	
1000002	A	G	HELD_FEM_EFF	3,35	0,3	12	21	3	22	25	19	
1000006	A	G	HELD_FEM_CC	2,77	0,36	31	58	4	22	31	13	
1000006	A	G	HELD_ALL_CC	2,34	0,43	44	82	6	38	58	18	

DAYSNP	ALLELE1	ALLELE2	COMPARISON	RR1	RR2	SIZE A	FREQ1 A	FREQ2 A	SIZE B	FREQ1 B	FREQ2 B
10000014	A	C	HELD_ALL_CC	1,69	0,59	45	83	7	39	64	14
10000014	A	C	HELD_FEM_CC	1,68	0,6	31	58	4	22	37	7
10000025	C	T	HELD_MAL_LIP	1,46	0,68	20	29	11	36	43	29

Claims

1. An isolated polynucleotide encoded by a phenotype associated (PA) gene; the polynucleotide is selected from the group comprising

5 SEQ ID 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 13, 14, 15, 16, 17, 18, 19, 20, 21,
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134, 135, 136, 137, 138, 139, 140, 141, 142, 143, 144, 145, 146, 147, 148,
149, 150, 151, 152, 153, 154, 155, 156, 157, 158, 159, 160, 161, 162, 163,
15 164, 165, 166, 167, 168 with allelic variation as indicated in the sequences
section contained in a functional surrounding like full length cDNA for PA
gene polypeptide and with or without the PA gene promoter sequence.

2. An expression vector containing one or more of the polynucleotides of claim
1.

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3. A host cell containing the expression vector of claim 2.

4. A substantially purified PA gene polypeptide encoded by a polynucleotide of
claim 1.

25

5. A method for producing a PA gene polypeptide, wherein the method
comprises the following steps:

30

- a) culturing the host cell of claim 3 under conditions suitable for the
expression of the PA gene polypeptide; and
- b) recovering the PA gene polypeptide from the host cell culture.

6. A method for the detection of a polynucleotide of claim 1 or a PA gene polypeptide of claim 4 comprising the steps of:

contacting a biological sample with a reagent which specifically interacts with the polynucleotide or the PA gene polypeptide.

5

7. A method of screening for agents which regulate the activity of a PA gene comprising the steps of:

contacting a test compound with a PA gene polypeptide encoded by any polynucleotide of claim 1; and detecting PA gene activity of the polypeptide, wherein a test compound which increases the PA gene polypeptide activity is identified as a potential therapeutic agent for increasing the activity of the PA gene polypeptide and wherein a test compound which decreases the PA activity of the polypeptide is identified as a potential therapeutic agent for decreasing the activity of the PA gene polypeptide.

10

8. A reagent that modulates the activity of a PA polypeptide or a polynucleotide wherein said reagent is identified by the method of the claim 7.

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9. A pharmaceutical composition, comprising:

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the expression vector of claim 2 or the reagent of claim 8 and a pharmaceutically acceptable carrier.

10. Use of the reagent according to claim 8 for the preparation of a medicament.

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11. A method for determining whether a human subject has, or is at risk of developing a cardiovascular disease, comprising determining the identity of nucleotide variations as indicated in the sequences section of SEQ ID 1-168 of the PA gene locus of the subject and where the SNP class of the SNP is "CVD" as can be seen from table 3; whereas a "risk" genotype has a risk ratio of greater than 1 as can be seen from table 6.

30

12. A method for determining a patient's individual response to statin therapy, including drug efficacy and adverse drug reactions, comprising determining the identity of nucleotide variations as indicated in the sequences section of SEQ ID 1-168 of the PA gene locus of the subject and where the SNP class of the SNP is "ADR", "EFF" or both as can be seen from table 3; whereas the probability for such response can be seen from table 6.

5

13. Use of the method according to claim 12 for the preparation of a medicament tailored to suit a patient's individual response to statin therapy.

10

14. A kit for assessing cardiovascular status or statin response, said kit comprising
a) sequence determination primers and
b) sequence determination reagents

15

wherein said primers are selected from the group comprising primers that hybridize to polymorphic positions in human PA genes according to claim 1; and primers that hybridize immediately adjacent to polymorphic positions in human PA genes according to claim 1.

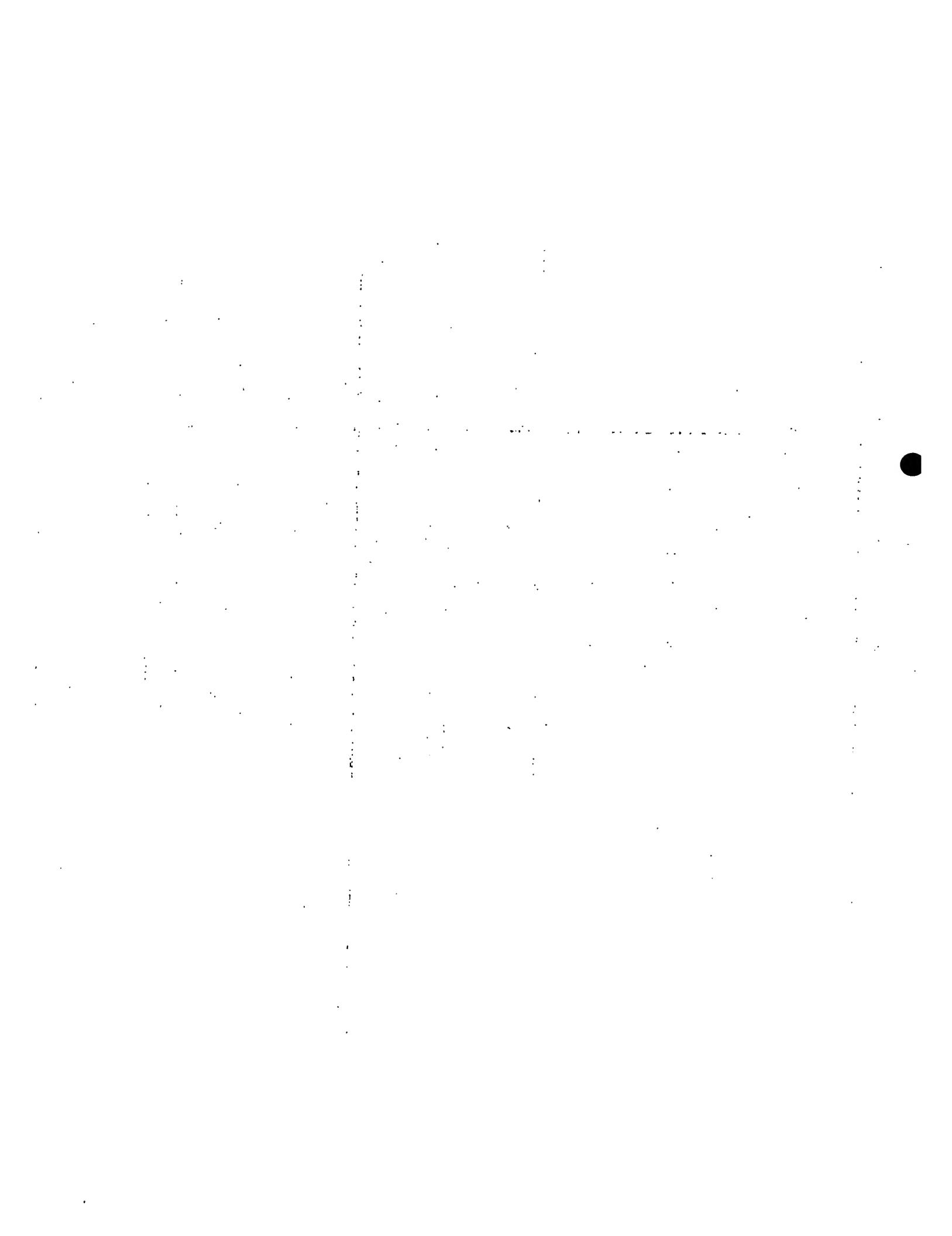
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15. A kit as defined in claims 12 detecting a combination of two or more, up to all, polymorphic sites selected from the groups of sequences as defined in claim 1.

25

16. A kit for assessing cardiovascular status or statin response, said kit comprising one or more antibodies specific for a polymorphic position defined in claim 1 within the human PA gene polypeptides and combinations of any of the foregoing.

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Single Nucle tide Polymorphisms sensitively predicting Advsverse Drug Reactions (ADR) and Drug Efficacy

Abstract

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67
19. Aug. 2002

The invention provides diagnostic methods and kits including oligo and/or polynucleotides or derivatives, including as well antibodies determining whether a human subject is at risk of getting adverse drug reaction after statin therapy or whether the human subject is a high or low responder or a good a or bad metabolizer of statins. The invention provides further diagnostic methods and kits including antibodies determining whether a human subject is at risk for a cardiovascular disease. Still further the invention provides polymorphic sequences and other genes.

The present invention further relates to isolated polynucleotides encoding a phenotype associated (PA) gene polypeptide useful in methods to identify therapeutic agents and useful for preparation of a medicament to treat cardiovascular disease or influence drug response, the polynucleotide is selected from the group comprising:

SEQ ID 1-168 with allelic variation as indicated in the sequences section contained in a functional surrounding like full length cDNA for PA gene polypeptide and with or without the PA gene promoter sequence.

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67
19. Aug. 2002

Sequences:

SEQUENCE LISTING

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<211> 578

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (154)..(154)

<223> baySNP90_C154T

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gggtgacacc aaggagcagc gcattcctgaa ccaygtgctg cagcatgcgg agagccccggg. 180

aacgcacaaa gcgtgctgga ggccatttgc acctactgcg agcagaagga gtggggccatg. 240

aacgtggcg acaagaaaagg tggggtccgg gccagcaggt gtcagctct gggacaggga. 300

cccaggacca ggcattcaaag cccttacagg agaagctgtt atcacccctt ttcagggggg. 360

ctgggaaccc tgggatatgc ccagataggg ctggggggct cctctggagt cccagggtgc. 420

cagggtcctt gatgaccctt gcaggccctg ctgcctgctg ccccaagaca acaggcccc. 480

acactcacag ggtctgacgg tggtgcaagg ccccttgaac tctgttctgg gcaccatgga. 540

acctgcttgg ggaccagtca gacaggttctt cctgggcc. 578

<210> 7

<211> 224

<212> DNA

- 10 -

<213> Homo Sapiens

<220>

<221> variation

<222> (58) .. (58)

<223> baySNP99_C58T

<400> 7

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gagaaaagag caggagtccct gggctttccc agttgagact gctggagctg agacacagta 120

ctctcttaaa gaaggtgggg agctgcccag ggcagaaccc actggtgttc atgactaccc 180

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<211> 630

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (468) .. (468)

<223> baySNP140_C468T

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tacagggtca catggtatct acttatctgt ggctttatccc tctttgtccc catattctat 180
cccaattaca tagactcctt gtttatgcc tttataactt gagaaactgt ctcagatcct 240
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cataatattt tactatgtaa tttctcccat gattctgtat ttgtgttact tactttgagt 420
gtgtctctga cctgggcctt gataatagga cccaaaatcc catcttcyyt catattggga 480
ttcactgtat gtttggtgaa ggactcatct tcgtactgtg tgtacataac tttcttataa 540
tgtttccaa tttgggttga gaaattatcc aaatgctgag acctgtatcc tcttaaagtgc 600
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<210> 9

<211> 968

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (587) .. (587)

<223> baySNP152_A587G

<400> 9

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gagaaggtag	aaagaatgtc	tccaggTTT	gcaggaagaa	agctagaagg	gggagggggtg	480
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aagacccagg	ggaagggcct	gagcacagag	gtgggaggTT	ccagccrgcg	ccctgcgagc	600
tggctgaatt	acctccctct	cctgggaggg	tgacacacgc	tcttggtacc	attctccatt	660
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cacagggttg	gtcaggaagt	aaaagaagac	tgtgaatgtg	aaatgcttcc	aaaatataaa	780
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gcaccaggga	ctggtttcag	ggaagacaat	ttttccatgg	acagggaggt	ggggatggtt	900
tcagaatgaa	actgttccac	cccagatcat	caggcattag	tttgatcctc	ataaggagtg	960
aacaacct						968

<210> 10

<211> 709

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (209)..(209)

<223> baySNP214_A209G

<400> 10

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gggctgccat ggtgacggcg gtggcctacc gcttggccga gcagcacccgg cagatagagg 180

agacctggc tcatttccac ctcaccaarg acatgctgct ggaggtgaag aagaggatgc 240

gggcccagat ggagctgggg ctgaggaagc agacgcacaa caatgccgtg gttaagatgc 300

tgccttcctt cgtccggaga actcccgacg ggaccggtga gggcctgctg ggggctgaca 360

tgcctgtcct gctcctgcca ggaactgagc cgcagtgggg gaggttggat tcgcccagtgg 420

atgccttcac actggtgatg cacgatttg ccagagctag aaactccctc acatctctgt 480

tttccatttt tattaactga tttgtcatgt gtgtgactca gggcagtttta atttcctgg 540

tcagacctcc caagcaagtg ctatctctaa aaccacagcc ctgctcagaa cagtgcctt 600

agctggcaa ggtgacacac gcctgtcatc cagctactta aaaggctgag gtgggaggac 660

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<210> 11

<211> 674

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (339)..(339)

<223> baySNP221_C339G

<400> 11

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ctttaacaa agcacctaaa atcttttaa tgccataatc ttttgacaaa atgccacctg 180

atgatattcc agcttctgag cctccagttg atcatgctga cgttgttagct ctttttttg 240

tttctgaagc tcatctgcct tcttcataag ttcattttct tgtaaagcaa taatattttc 300

atattctttt agttcttcct ctgtgaagaa ctgttgctsa tctaattgtct aaaagaaaaa 360

aaatgaggaa agaataatct caaaactaca acaataaagt ctgatttgc tttaaaattc 420

- 15 -

tatggtagc aaagtataaa agagtatcta taggattacg cctttggagg taagaaggaa .. 480
ggggaaaataa gtaatatata tttatgtact catttgaaca aaaggaaaaa caggaagaaa 540
aactcaagag attctcttct gggattgaca actaaaggca aagagtgaga actgggtaga .. 600
agaggcaaag agaacgtggg aagagaaaata aaatttgtca ttccccccaa ccctcactga .. 660
aacatgaaat ttta .. 674

<210> 12

<211> 811

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (189)..(189)

<223> baySNP224_C189T

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catgtgtttg gttctggggc cccttttac tcccccttgag ctgaaaggaa cccctttgg	720
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<210> 13

<211> 776

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (465)..(465)

<223> baySNP294_C465T

<400> 13

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gggagagcac agcttgaatg gcgcacgtaca tggcaggac attgaagggtt tcaaacatga	180
tcttaaagga caatgaagaa taaaagatta tttggcaaaa atcacagcac ttcttggcct	240
gtagaatggt ttgtaatctc attaaggca ggaactatgc cagtttgata gttcattatt	300
ccacacccac tcccccaggg cctggcatgg accacagaca tagcaggtcc tcagagagtt	360
ctgttgcact gatgtttgaa ggggagaaag tatctggcca agcatccatg ttgcatttt	420
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ttgttagaagg agtggtgcca gatctagtgg agacaaaaagc catatgagcc tggcagactc	600
ccaatggctg gaaaattctc attgacattc ccatgctggg agagtgtggc accatgttac	660
acaggatggg atggatctgg agaggagaat ggtcagccct ggaaatttctt gattgacatt	720
taaagggtat actccttctc acagggcccc agtaggactc agactggaac agtatt	776

<210> 14

<211> 1920

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (215) .. (215)

<223> baySNP307_C215T

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agggctgaga gcaaaaaccca gggtcttgtc tggattccca ggccagttac tgccctcttg	180
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caccatgaag acgaaagaga aaatcgctca catggcagt gtagaacggg cgccccgtga	300
tggttcaggg atgaccctcc tctgttagctg cccagaggcc aacaccgccc ataccatagc	360
cactgtcccc aagtcagcct ggagggaaaga gagcaggtca cactcacctg attctgtatga	420
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ggtcagcagg gcccagagtc agggtcctcc gctgcctgag gcgtcaacat gcctgcctgc	660
aatgtgtttg tgcacatgctg tgcacatgtg tatgtggata aacatgtctg tgcacgtgtg	720
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cactcaggaa ggcttctcac tggaggaagg gcccagttca ggacacacag ggacatctcc	1740
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gactgtccca atgacagccc tagcaagaag agacaagaaa caagacaagt tcacgttgtc	1860
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<210> 15

<211> 587

<212> DNA

<213> Homo Sapiens

- 20 -

<220>

<221> variation

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<223> baySNP411_A397T

<400> 15

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gccttcctcc cccaggcatg gctggggact ggaagaccac cttcaccgtg gcgcagaatg 120

agcgcttcga tgcggactat gcggagaaga tggcaggctg cagcctcagc ttccgctctg 180

agctgtgaga ggggttcctg gagtcactgc agagggagtg tgcgaatcaa gcctgaccaa 240

gaggctccag aataaaagtat gatttgtt caatgcagag tctctattcc aagccaagag 300

aaacctcgag ctgaaagagt gatcgccac tggggccaaa tacggccacc tccccgctcc 360

agctcctcwa cttgccctgt ttggagaggg gagagggctt ggagaagtaa aaccaggag 420

acgagtagag gggaatgtg tttaatccca gcacgtcctc tgctgtcctg ccctgtgtcg 480

ttggggatg gcgagtctgc caggcggcat cacttttct tgggttcctt acaagccacc 540

acgtatctct gagccacatt gaggggaggg gaatagccat ctgcata 587

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<212> DNA

<213> Homo Sapiens

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<222> (323)..(323)

<223> baySNP449_C323G

<400> 16

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tgttcttata ctctactgtt tccacgttaa aaccagtggt aggaatggtg gtgactatct 180
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<210> 17

<211> 844

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (123)..(123)

<223> baySNP466_C123T

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ggyctggtgt ggcttctccc accatcaactg tggcgatgcc tggtgtccct gcctttctcc	180
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ccttagtgaca tgtagggga agggggccctg ggaattcatt gactctgaac ctttcatttt	360
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<210> 18

<211> 561

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (497)..(497)

<223> baySNP472_A497G

<400> 18

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aagtgttaggg ccaggaagca taagtgggta actcagaaaaa gctgtggcca ctgacactgg 180

ggctgcggcc aactgacgtc tgctctgctc ttgcatacttta catcctttgt tttggagttc 240

cctgcatactt acatcctttg ttttgagct ccccactggg tatgggagtc tcagaaccca 300

cagggattaa ggagacaagt tttctctcca acttacaagg gatctcaattt acccatcata 360

cttcttttcc tggggtccgc caatatgggg cctccctaca gaacgctttt gatgtgctgg 420

gcttcacttc agaggagaaaa aactccatgt ataagctgac aggccatc atgcactttg 480

gaaacatgaa gttcaarctg aagcagcggg aggagcaggc ggagccagac ggcactgaag 540

gtgggaggca ggattcttgg g 561

<210> 19

<211> 832

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (402)..(402)

<223> baysNP542_A402G

<400> 19

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aaagtgttgc tgacaaggtg aaatacataa tttcactttc tgagtactta agaacagtta 180

gctgttatta tcaagcaaat ttgttatcaag tggccaca cattcagcca tggctgtg 240

gtactgcctg gttgtatag gctgctggac aggcagaggc tccttcttcc taaaagccct 300

tggcagtcac attatattta cctgtttctt ggtattaagc cgtaatttgc atgattgagc 360

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aacatgttct gaaagcgtgt catgaacacc atgtcccatg ggttagccgaa gtcaaagatt 480

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- 25 -

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tggttcttct aacttctatt ttaaagttaa tccttttag attaggattc agagcatcta 720
gagtattaaa atgccttaag acatattatt gatacttatt atctgccagc cttagtaaa 780
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<210> 20

<211> 988

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (87)..(87)

<223> baySNP739_C87G

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tcattcagca aatatcctcc gatgattcac atgttaggttt cagaaagcct gaaaaaaatg 240
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cctgaggccc accaataaca aaatctgcc	360
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tcaaggatt tcgcaggcac aactgctttg atgactttct cctttagggc atccctcatt	420
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cctaagaaag aagtcaccat gtgatcaagc tcgatttatg ttgagagatt aaacagctt	900
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<210> 21

<211> 851

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (140)..(140)

<223> baySNP821_A140C

<400> 21

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tgaaacagcc	ttctatttggaa	aaaagatgcc	atctggactt	tggatagaga	ggataagtca	780
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<210> 22

<211> 880

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (257)..(257)

<223> baySNP1005_A257G

<400> 22

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cacagccctt gtctaccatt gctagcctca ggcaaattgac tccacttctc tatgcctcag	240
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aaggtttaaa ggacacatgc tgggtatatg tagggctgca gacatgagga gtcctggacg	360
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<210> 23

<211> 550

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (287)..(287)

<223> baySNP1055_A287T

<400> 23

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- 30 -

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<210> 24

<211> 668

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (354)..(354)

<223> baySNP1056_A354G

<400> 24

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- 31 -

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cacaggggga ccccaggcag agccctgccc catgctcggc ctcagttacc tcatacgaa	600
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<210> 25

<211> 789

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (257)..(257)

<223> baySNP1085_A257G

<400> 25

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gcaggctgcc caccaggggc agggacagga ggctcttggg gtacttggca ccagggcacc	180

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<210> 26

<211> 789

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (104)..(104)

<223> baySNP1086_A104G

<400> 26

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<210> 27

<211> 1756

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (1687)..(1687)

<223> baySNP1092_C1687G

<400> 27

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gcccattccc tcttagcca gagccggggt gtgcagacgg cagtcactag gggcgctcg	720
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cagagagaag tcgaggaaga gagagacggg gtcagagaga ggcgcgggc gtgcgagcag	1680
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<210> 28

<211> 565

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (454) .. (454)

<223> baySNP1096_G454T

<400> 28

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taccagtctg ctccgttcct gaggacccta actttctca gcggaagcag gaagacctga	180
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atggagctgt tcccaggctg cctgtgcaca ggctggagag gagggttaca tcacttggcc	540
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<210> 29

<211> 842

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (249) .. (249)

<223> baySNP1101_C249T

<400> 29

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tg 842

<210> 30

<211> 656

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (307) .. (307)

<223> baySNP1204_A307G

<400> 30

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aacggctccc cctgggtgt gtactgaggg gcagaaggga ggtgacgtgg gagtcagggg 180

tcaagtgtccc agccctgccc ccaacccttt gggcaagctc ttgcgtctgt ttccccatct 240

agcgcatgag gacccaactc cttgcccgtt aagcatctgg aattgtcatg agagccaaaa 300

ctaattrtaa tgtgagtgcc cttgctaaag atcaaagact gagccatgca cgcaagtcatc 360

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<210> 31

<211> 615

<212> DNA

<213> Homo Sapiens

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<221> variation

<222> (180)..(180)

<223> baySNP1504_C180T

<400> 31

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- 40 -

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<210> 32

<211> 527

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (153) .. (153)

<223> baySNP1511_G153T

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tcttggatat tcattttggg actctgcgtc aakgaacaat gagcgctgca ccctaataaca	180
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gaatgcctct catgctaaga agggatgccc aggacatggg aatgccactc cccacaagtc 480
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<210> 33

<211> 663

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (284)..(284)

<223> baySNP1524_A284C

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<210> 34

<211> 609

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (367)..(367)

<223> baySNP1556_C367G

<400> 34

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<210> 35

<211> 740

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (699)..(699)

<223> baySNP1561_A699C

<400> 35
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<211> 1181

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<213> Homo. Sapiens

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<222> (389) .. (389)

<223> baySNP1582_C389T

<400> 36

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- 46 -

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<211> 531

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (443) .. (443)

<223> baySNP1638_A443G

<400> 37

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gtgggcagga gatatgttc cgccatggag acatcctggc cctctttccc tacctctcag 180

tgcacatgga ccctgacatc caccctgagc ccaccgtctt caagtacgat cgcttcctca 240

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tgaagctctt tatcctgctt atggtcacac actttgactt agagttggtg gaccctgaca 420

caccactacc ccatgttgac ccrcagcgct ggggtttgg caccatgcag cccagccacg 480

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<211> 711

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (648)..(648)

<223> baySNP1653_G648T

<400> 38

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ctaaatatata tcaatcta atttgaggac acatggaa atatttatga ttcatctgct 180

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cagccgactc agggtgttcc tgtggactga acaatctctt gctgctgtac tggagggcct 360

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taaatgagat tcctggtaa aaaggaccag agcagtcatt ttacagttga ggaaactgtt 480

gctctgagaa gtgaggatt tattcatgac tacactgatg gtgagtgccc atgtcaggc	540
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ctgATGGGAG gtcCCAAAGC gggAGGAAGA aggAAAGTCT tgCCACKGC atTCCTCAG	660
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<211> 2438

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (398)..(398)

<223> baySNP1662_C398T

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cTTATGCTTA tattaAGTTG tagCATATGc attTACCGAT agttaACCgt attAACCTAC	300
agaaaATGTC caggAAATG gtctATTCT tattCTATTt ttGACCTAAA gaaaATCTT	360

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- 50 -

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<213> Homo Sapiens

<220>

<221> variation

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<211> 813

<212> DNA

<213> Homo Sapiens

<220>

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<222> (89)..(89)

<223> baySNP1722_C89T

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<210> 42

<211> 524

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (210)..(210)

<223> baySNP1757_A210G

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cgtggatgga agaggcgagg gcaaaaggaa gagatgaggg gcatggagag agactcaggg 360

aagaaggaga aagagcaatc atgcagcttg ggacaaatct tttgttgctt tatcagattc 420

tcagtcaatc aattgggtgtt tgctagaacc ggggtacgca ggggagtgtt qaaqagagaa 480

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<210> 43

<211> 1279

<212> DNA

<213> Homo Sapiens

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<222> (240) .. (240)

<223> baySNP1765 A240G

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[View Details](#) | [Edit](#) | [Delete](#)

300

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<211> 359

<212> DNA

<213> Homo Sapiens

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<221> variation

<222> (200)..(200)

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ccaaagggtt cccaacaccr tctctcctct gatttctgtg acaaatgtgg aaagctactt 240

gcttgagggt actgggggaa ctgatggggg aactttcatc ctgttaggaa ctccgcttcc 300

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<210> 45

<211> 437

<212> DNA

<213> Homo Sapiens

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<222> (127) .. (127)

<223> baySNP1799_A127G

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caagaaaagtt ctgagcactg aggaagcaat gctgaataaa atatcccatg ccctcagcaa 360

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<210> 46

<211> 752

<212> DNA

<213> Homo Sapiens

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<223> baySNP1806_A201G

<400> 46

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<210> 47

<211> 527

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (413)..(413)

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<210> 48

<211> 600

<212> DNA

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<222> (323) .. (323)

<223> baySNP1870_C323T

<400> 48

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<211> 725

<212> DNA

<213> Homo Sapiens

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<221> variation

<222> (115)..(115)

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cccg	725

<210> 50

<211> 493

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (214)..(214)

<223> baySNP1988_C214T

<400> 50

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gaccCACCC tgcacacagg gtgcccaagtg tccttccgc tcctctccca ggccctctaca 360

actactacga cgacgagaag gaaaagctgc aaatcgatgg aatgcccctg gcccacaagc 420

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tgctaaccaa aga 493

<210> 51

<211> 1161

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (349) .. (349)

<223> baySNP2000_C349T

<400> 51

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tctttcccc acaacaatat tcatgtgcct agaccaagaa gttattatct cgaactgaca 180

ctgagccaca acccaaacaa cccagctctc cctaagcttc aaactagact acttctccat 240

aatattcattc cctgttagcat tggcggttac atggtccatc atagaattct cactgtgata 300

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<210> .52.

<211> .2915

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (338) .. (338)

<223> baySNP2071_A338G

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<211> 1557

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (876) .. (876)

<223> baySNP2078_G876T

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<211> 503

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (415)..(415)

<223> baySNP2085_G415T

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- 70 -

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<210> 55

<211> 553

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (183) .. (183)

<223> baySNP2095_C183T

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cataatacac aattccagcc agcaagctgt ccccaggatg cataatacac aattccagcc 420
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cctaattata acc 553

<210> 56

<211> 275

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (67)..(67)

<223> baySNP2119_A67G

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<210> 57

<211> 393

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (176)..(176)

<223> baySNP2141_A176G

<400> 57

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agaagctgta gcatcggtag gttccgctgt gggcggcggt caccgtatgt atgggraaac 180

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caaattggtc aaagccatac cgagtctgac actgttagggt tacgtcccct cctgacgaca 300

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<210> 58

<211> 423

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (318) .. (318)

<223> baySNP2182_A318G

<400> 58

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aggcgggcca agggtgarga atctatcctg aaagtagcaa gaagagtgaa catttaacca . 360

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gac . 423

<210> 59

<211> 542

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (296) .. (296)

<223> baySNP2234_G296T

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<210> 60

<211> 859

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (227) .. (227)

<223> baySNP2281_A227C

<400> 60

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<211> 371

<212> DNA

<213> Homo Sapiens

<220>

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<222> (77)..(77)

<223> baySNP2298_A77C

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<210> 62

<211> 825

<212> DNA

<213> Homo Sapiens

<220>

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<222> (286) .. (286)

<223> baySNP2341_C286T

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<210> 63

<211> 565

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (165)..(165)

<223> baySNP2357_A165G

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<210> 64

<211> 549

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (50)..(50)

<223> baySNP2366_G50T

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<210> 65

<211> 609

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (170) .. (170)

<223> baySNP2423_A170G

<400> 65
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<211> 636

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<213> Homo Sapiens

<220>

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<400> 66

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<220>

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<222> (299) .. (299)

<223> baySNP2995_A299C

<400> 67

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<211> 934

<212> DNA

<213> Homo Sapiens

<220>

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<222> (777) .. (777)

<223> baySNP3360_G777T

<400> 68

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<211> 372

<212> DNA

<213> Homo Sapiens

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<221> variation

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<223> baySNP3464_A110G

<400> 69

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<223> baySNP3689_C298G

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<212> DNA

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<223> baysNP3975_A65C

<400> 71

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<222> (239) .. (239)

<223> baySNP3976_A239G

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<223> baySNP4206_A304T

<400> 73

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<222> (583)..(583)

<223> baySNP4838_A583G

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<211> 921

<212> DNA

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<222> (74)..(74)

<223> baySNP4912_A74G

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<210> 79

<211> 486

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<222> (366) .. (366)

<223> baySNP5296_A366G

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cgtccrggga ggttttctcc ataaacttgg tggaaggca gtgggcaaat ccaggagcca 420

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<210> 80

<211> 486

- 100 -

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (172)..(172)

<223> baySNP5298_C172T

<400> 80

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- 101 -

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (101)..(101)

<223> baySNP5320_A101G

<400> 81

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gagtagtagt gattaagttg gccagtgccc agaagtgcctt actgggtgcc aggattgttg 180

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<210> 82

<211> 1975

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (1604)..(1604)

<223> baySNP5361_A1604C

<400> 82

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<210> 83

<211> 527

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (134)..(134)

<223> baySNP5457_A134G

<400> 83

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gccattttag agttccagc agaagatgac cttccccctt gtgcgctgga aacggggcac 360

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<210> 84

<211> 562

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (61)..(61)

<223> baySNP5704_C61T

<400> 84

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<210> 85

<211> 2246

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (1960) .. (1960)

<223> baySNP5717_A1960G

<400> 85

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<211> 564

<212> DNA

<213> Homo Sapiens

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<221> variation

<222> (71)..(71)

<223> baysNP5959_A71G

<400> 86

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- 109 -

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<210> 87

<211> 494

<212> DNA

<213> Homo Sapiens

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<222> (340)..(340)

<223> baySNP6162-C340G

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- 110 -

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<210> 88

<211> 1898

<212> DNA

<213> Homo Sapiens

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<222> (906) .. (906)

<223> baySNP6236_C906T

<400> 88

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<211> 1351

<212> DNA

<213> Homo Sapiens

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<221> variation

<222> (151)..(151)

<223> baySNP6482_A151G

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- 114 -

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<211> 851

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (289) .. (289)

<223> baySNP6498_A289G

<400> 90
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<210> 91

<211> 2958

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (348) .. (348)

<223> baySNP6744_C384T

<400> 91

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<210> 92

<211> 689

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (63)..(63)

<223> baysNP7133_C63G

<400> 92

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<210> 93

<211> 2184

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (1603)..(1603)

<223> baySNP8021_A1603G

<400> 93	
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ggCTGGGTGA CCTCTGACTC CTGGGTTCAA GTGATTCTCC TGCCTCAGCC TCCTGAGTAG	240
CTGGGATTAC AGGCGCCCAGC CATCATGCC GGCATAATTTC TGTATTTCA GTAGAGATGG	300
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CGGTCTCCCA AAGTAGTGGG ATTACAGGCG TGAGCCACCG CACCCGGCCT GGAACAGGCC	420
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CAGTGAGAAG TGAACAAGTA GGGCCGGGCT CACGGCTGTA ATCCCAGCAC TTTGGGAGGC	720
TGAGGTGGGT GGATCACCTG AAGGTCAGGA GTTCAGAGACC ATTCTGACCA ACATGGAGAA	780
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<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (118)..(118)

<223> baySNP8060_A118G

<400> 94

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gaacccagac agagtctcta tgggttgagg aaatagagtt tggacttcgg agaagctcaa	540
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<210> 95

<211> 1151

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (255) .. (255)

<223> bySNP8210_A255G

<400> 95

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ctttaaagaa gaatgttaat attttacttt tacagtcatt ttcctacatc ggaatccaag 180

ctaatttcta atggccttcc ataataattc tgcttagat gtgtatacag aaaatgaaag 240

aaactagggt ccatrtgagg gaaaacccaa tgtcaagtgg cagctcagcc accactcagt 300

gcttctctgt gcaggagcca gtcctgatta atatgtggg attagtgaga catcagggag 360

taagtgacac tttgaactcc tcaaggacag agaactgtct ttcatttttg aaccctcggt 420

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cagatttcaa aagaacagaa ggactgaaga ccagctgtgt ttcttaacta aatttgcctt 540

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<210> 96

<211> 518

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (46) .. (46)

<223> baySNP8592_C46T

- 125 -

<400> 96
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tcaatgaccc ctgcattgac ctcaacaaca tagtctacat gatcttagtat gattccaccc 180
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caccaactgc ttagcacccc tggccaaggt catccatg 518

<210> 97

<211> 538

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (181)..(181)

<223> baysNP8816_C181G

<400> 97

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cagcaccagg tcccggaggg acagatgagg gatccctagt aaacagctcg tggacgcact	480
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<210> 98

<211> 524

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (196) .. (196)

<223> baySNP8846_A196G

<400> 98

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ctgtcgccctg acagcataacc tcaaaaaggcc ctagctaata aacctgtaag cccagcattt	120
gagagaaggt agggtgtgtat tatataaaag gttgtgtaca actccacgag gtgaaaaata	180
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tatattaaac cttcagatta atgactggct acagagtaac aaaaaataaa gaatttaatg	300
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ctgttgtat tgataagtaa agccactcat tgaaaaaccc aattccaaac accacagttt	480
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<210> 99

<211> 1150

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (351)..(351)

<223> baySNP8943_A351C

<400> 99

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<211> 783

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (88)..(88)

<223> baySNP9193_C88G

<400> 100

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ggtcgtgaccg tagacctgcg cgttggcgcc ggccgcggcg ttgaactcgt aggccggcgcc 180

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- 130 -

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<210> 101

<211> 611

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (329) .. (329)

<223> baySNP9443_C329T

<220>

<221> misc_feature

<222> (391) .. (391)

<223> Unsure

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<210> 102

<211> 589

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (187)..(187)

<223> baySNP9516_A187G

<400> 102

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<210> 103

<211> 1584

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (1546) .. (1546)

<223> baySNP9698_A1546G

<400> 103

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<211> 744

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (277) .. (277)

<223> baysNP9849_C277T

<400> 104

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<210> 105

<211> 560

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (249) .. (249)

<223> baySNP9883_A249G

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<222> (506) .. (506)

<223> Unsure

<400> 105

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<211> 555

<212> DNA

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<221> variation

<222> (249) .. (249)

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<211> 609

<212> DNA

<213> Homo Sapiens

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<221> variation

<222> (107)..(107)

<223> baySNP10481_A107T

<400> 107

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<211> 2388

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (367)..(367)

<223> baySNP10542_C367T

<400> 108

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<211> 597

<212> DNA

<213> Homo Sapiens

<220>

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<223> baySNP10600_A510G

<400> 109

- 142 -

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<211> 569

<212> DNA

<213> Homo Sapiens

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<222> (402)..(402)

<223> baySNP10621_C402T

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<210> 111

<211> 631

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (431) .. (431)

<223> baysNP10745_A431G

<400> 111

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<210> 112

<211> 2956

<212> DNA

<213> Homo Sapiens

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<221> variation

<222> (2831) .. (2831)

<223> baySNP10747_C2831T

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<211> 557

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (64) .. (64)

<223> baySNP10771_C64G

<400> 113

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<210> 114

<211> 544

<212> DNA

<213> Homo Sapiens

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- 149 -

<221> variation

<222> (339) .. (339)

<223> baySNP10870_A339G

<400> 114

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<222> (1290)..(1290)

<223> baySNP10877_A1290C

<400> 115

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<211> 623

<212> DNA

<213> Homo Sapiens

<220>

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<222> (140)..(140)

<223> baySNP10948_G140T

<400> 116

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<210> 117

<211> 555

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (286) .. (286)

<223> baySNP11001_C286T

<400> 117

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<210> 118

<211> 1051

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (215)..(215)

<223> baySNP11073_C215G

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<211> 478

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (116)..(116)

<223> baySNP11153_C116T

<400> 119

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tctaccctat cttggcgct atgcccagta attctaggca ggcagtcatt cttagaggag 180

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tgttatcttt cagcagctt a gttaaggca gccagcattc tcatacgtagg aatggaaagc 420

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<211> 1197

<212> DNA

<213> Homo Sapiens

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<222> (194)..(194)

<223> baysNP11210_C194T

<400> 120

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aggagaaaagg agcaggagtc ttggccaaaa taaagatttgg gttttgttc cctaagccag 180

gaaatgacat gatyagattt gcttttaagg atccttccaa actgctaaag acatgttagat 240

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acatggtaaa actgcagtct ctactaaaaaa tacaaaaatt agccagtcgt ggtggcgcgc 540

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<210> 121

<211> 500

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (225)..(225)

<223> baySNP11248_C225T

<400> 121
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<210> 122

<211> 2955

<212> DNA

<213> Homo Sapiens

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<221> variation

<222> (1110)..(1110)

<223> baySNP11372_A1110G

<400> 122

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<211> 1004

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (486)..(486)

<223> baySNP11449_C486G

<400> 123

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atctcttaag actgtatggtt tagaatatacg cagaccatat ttttatgtcg ctttagactc 180

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<210> 124

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<212> DNA

<213> Homo Sapiens

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<222> (489)..(489)

<223> baySNP11450_A489T

<400> 124

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accagcctgg tcaacacggt gaaacccct ctctactaaa aata 1004

<210> 125

<211> 540

<212> DNA

<213> Homo Sapiens

<220>

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<222> (511)..(511)

<223> baySNP11470_C511T

<400> 125

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agaaaaacgag tttgtcagta aatgttagtaa ttaaggaagt cccaaagtctg tatttttaag 180

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cctccccccg ccaccccta ccaaaaccat accttagtca ctttggatt ataaccgtct 300

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<210> 126

<211> 641

<212> DNA

<213> Homo Sapiens

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<221> variation

<222> (556)..(556)

<223> baySNP11472_A556T

<400> 126

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GTGTCATGTA AAAATACATT AACTAGTAAC TTGTCAAGAA GTGCTAGATT CCATTCTGT 420

GCGGGTCCAT TTATATATGT TCCTGAACAG CTAATATTC ATTAGTTTC CAATTAAACT 480

- 166 -

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<212> DNA

<213> Homo Sapiens

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<222> (485) .. (485)

<223> baySNP11487 A485T

52202

<221> misc feature

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<223> N region

<400> 127

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- 168 -

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<210> 128

<211> 1547

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (533)..(533)

<223> baySNP11488_C533G

<220>

<221> misc_feature

<222> (848)..(947)

<223> Unsure

<400> 128

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- 170 -

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<211> 799

<212> DNA

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<220>

<221> variation

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<223> baySNP11493_A171G

<400> 129

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<210> 130

<211> 535

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (455) .. (455)

<223> baySNP11502_C455T

<400> 130

- 172 -

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<210> 131

<211> 630

<212> DNA

<213> Homo Sapiens

<220> ..

<221> variation

<222> (258) .. (258)

<223> baySNP11534_G258T

<400> 131

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<210> 132

<211> 1854

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (274)..(274)

<223> baySNP11537_A274G

<400> 132

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<210> 133

<211> 588

<212> DNA

<213> Homo Sapiens

<220>

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<222> (185)..(185)

<223> baySNP11560_A185G

<400> 133

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<210> 134

<211> 620

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (353) .. (353)

<223> baySNP11578_C353T

<400> 134

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<210> 135

<211> 501

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (328) .. (328)

<223> baySNP11594_C328T

<400> 135

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gaaaaccaag agctggccac caccctgcca gcagtggtga cgggaatcat gccagtgact 240

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ctactggtat tatgttaactt t 501

<210> 136

<211> 1395

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (912)..(912)

<223> baySNP11624_C912T

<400> 136

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- 180 -

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<210> 137

<211> 572

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (277) .. (277)

<223> baySNP11627_C277T

<400> 137

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<211> 558

<212> DNA

<213> Homo Sapiens

<220>

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<222> (419) .. (419)

<223> baySNP11644_A419G

<400> 138

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<210> 139

<211> 597

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (146)..(146)

<223> baySNP11650_A146G

<400> 139

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cctcacgctg gccatgctct ggggrcttt tagacatttg gccaggggt ggcagaacta 180
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<210> 140

<211> 501

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (402) .. (402)

<223> baySNP11654_A402G

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<210> 141

<211> 501

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (331) .. (331)

<223> baySNP11655_A331C

<400> 141

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ttacttcaaa tatttctgca ctaaagctgt matattacaa gtttgttttc ctattagagg	360
tttccatttc catccattta tctttactt gaaaggacac cgtattttc acctaattccc	420
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<210> 142

<211> 1224

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (897) .. (897)

<223> baySNP11656_C897T

<400> 142

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- 187 -

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<210> 143

<211> 608

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (277) .. (277)

<223> baySNP11825_A277G

<400> 143

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atataaacacc aattgtggaa tgcaccacaa tgtcagcatt gtcctctgac ttgacatgaa 180

cattaaacttg taatgtgacc ttggaaataa tggaggtcaa tatataaaata ttcttacaaa 240

tcattaagga aaatgctaac aacccagcaa aaatggrrcaa aagatataaa taaaaagttc 300

acaagaaaaga aaatgcaaat ggttcttaat gtcttaaaac aatctcaata tcactcagag 360

tacaaataca aattaaaact actataagat acaattttca cttatcaggc tggcaaaggc 420

aattttcgtc cctgtctatt caaagtaaca ccattttctt tattagttcc agtgggcaag 480

taagagcaag gagtccacat agcggtggcc aaccaaagtg atgaaaggaa ggcacat 540
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<210> 144

<211> 574

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (246) .. (246)

<223> baySNP11914_A246T

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acacattatc ttcttcatt tttaaagagt tttttctggc atttttacaa ggcgttacta 360

- 189 -

gttggccct tgtaaccc tcatttcac cgtgattctg acattgagat gttcaaccag 420
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attaaaatca ttatcctcc ctttattaag gttactgttt aaaaaggtaa aggccctgt 540
gggagggaaat gtccgcatga tgctgtctgg aggg 574

<210> 145

<211> 569

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (449) .. (449)

<223> baySNP12008_C449T

<400> 145

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ataagcacat tacatcatcc tgcggtaacgg tgtctttcat accggtgatc attgtggtcg 300
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- 190 -

ttttaaagca gagcatcctc cacacagtgc tctcccccagg agaagtctgc tgtaaaacat 420
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<210> 146

<211> 1619

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (411) .. (411)

<223> baySNP12097_A411G

<400> 146
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cagagtttgg ctcttttgt tggcagcatg tattaactca ttctcttatta caattccct	1560

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<210> 147

<211> 482

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (412) .. (412)

<223> baySNP12366_A412G

<400> 147

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ccgagggccc agatgggaa gatgttcctg tagctcgtagt aggagatggc acaggacttg 180

ttgaagaccc cagcaatgtt ttcctaaaag aacacagaga aataaacaca aaggcttcac 240

cagttagtgc agggctagtc cctgaactgg agccctggct gttggcttgg ctggcttgg 300

ggtagttagtgc cctggaaagg gctaattaag caagagcata ggaaagcaag tagtccatcc 360

cgcatttgc atggcaccat cagccccagct cagactcccc gccctgcagc araccctaag 420

ccagggtcac cagccccctca caaccacatc agcttccacc aactcagcct catcccagg 480

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<210> 148

<211> 624

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (25)..(25)

<223> baySNP12619_A25G

<400> 148
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<210> 149

<211> 648

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (585) .. (585)

<223> baySNP13025_A585C

<400> 149

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atatacacac aaataagaga aaggaagaaa gaaagagaaa ggaaaggggc taacataaat 180

ttaaaaaaca caaatgatga ttaggtgtcc ttctgtttaa aaaattaata ttttccttt 240

caaagaaaaac attcttaatt gaggattaag tcaatatata agtatcttgc atctgttgct 300

tataaataca ggagagtat catagaagga acataatgat cacatatttc aacttacagt 360

acagtagtcc ccccttatttc gcagtttgc tttctgaggt ttcagttacc tgcagtcaat 420

- 195 -

cacatttcaa aaatattgaa tattccagaa atcagcaatt cataagttt aattttcatg	480
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ctcatccttt tgtccagtgc atccatgctg taaatactac ctgtmcacca gttacttaat	600
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<210> 150

<211> 625

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (504)..(504)

<223> baySNP13191_A504G

<400> 150

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agacttttat taaggcctta agtgcttagat tcaagatggc tgcccccttt gttctgtggg	180
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tccagtgttc tatttggtgg actaagggtg accttgcagc cccttacagc ccagccaaga	240
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gagcttcact gtgaagggc agacatcttc attactattt tctttccaa aaactcatat	300
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aacttttgt gagtactgcc tcttctcctc attccacagt gaatttctcc ccacctggtg	360
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acacaaaacag ccttttccca ggtacttgtt acctggagcg agtggacgag cagcatgcc 420
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<210> 151

<211> 581

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (47)..(47)

<223> baySNP13937_A47C

<400> 151
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gctcctttg actaaaactga gttacctcat aatgttcatg gttcagcgtc tctatgtga	480
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<210> 152

<211> 751

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (371)..(371)

<223> baySNP900002_G371T

<400> 152

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120

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180

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240

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<210> 153

<211> 511

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (322) .. (322)

<223> baySNP900025_G322T

<400> 153

- 199 -

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aagacatata tactttttt attcccctga gccatggaa agtaagctgc aaacatcttgc	300
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agttacatta tcatacttaa gagatatagt attaatacaa tattatctaa catgtgatcc	420
ctattnaaat ttcattttt ttcttccccca gaacagttct ttatagatgt tttaattttt	480
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<210> 154

<211> 598

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (261)..(261)

<223> baySNP900032_C261T

<400> 154

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- 200 -

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<210> 155

<211> 1086

<212> DNA

<213> Homo Sapiens

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<221> variation

<222> (116) .. (116)

<223> baySNP900045_C116T

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<221> misc_feature

<222> (986)..(1085)

<223> N_region

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<221> misc_feature

<222> (986)..(1085)

<223> Unsure

<400> 155

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cagaacgcta cagggacagc atggcctgga tggccatgta catggctggg gtgttgaagg 300

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<210> 156

<211> 1864

<212> DNA

<213> Homo Sapiens

<220>

<221> variation

<222> (877) .. (877)

<223> baysNP900065 A877C

<400> 156

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cctacgcatt ctacacacat atcccaaaac ttataagtaa aacagaaaaa aaaagaatta	120
gttttataaa aatcaataaa ttatactcag tgggtttttt ttttttttt tgagacggag	180
tctcgctctg tcgcccaggc tggagtgcag tggcgggatc tcggctcact gcaagctccg	240
cctccccgggt tcacgccatt ctcctgcctc agcctccaa gtagctggg a tacaggcgc	300
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<223> baySNP900082_A375G

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<210> 158

<211> 708

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<222> (136) .. (136)

<223> baySNP900096_A136G

<400> 158

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<211> 790

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<222> (426) .. (426)

<223> baySNP900107_C426T

<400> 159

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<223> baySNP900121_G263T

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<222> (263) .. (263)

<223>

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tggaatccac ggatgaaacc acaggtatgt gccttacaat atacaatggg agttttcatt 240

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- 209 -

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<211> 101

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<222> (816)..(816)

<223> baySNP900078_A816G

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<222> (103)..(103)

<223> baySNP10000002_A103G

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<210> 164

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<213> Homo Sapiens

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<222> (107) .. (107)

<223> baySNP10000006_G107A

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<210> 165

<211> 399

<212> DNA

<213> Homo Sapiens

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<222> (153)..(153)

<223> baySNP10000014_A153C

<400> 165

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<223> baySNP10000025_C291T

<400> 166

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<210> 167

<211> 3877

<212> DNA

<213> Homo sapiens

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<211> 620

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<221> variation

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PCT Application

EP0309126

